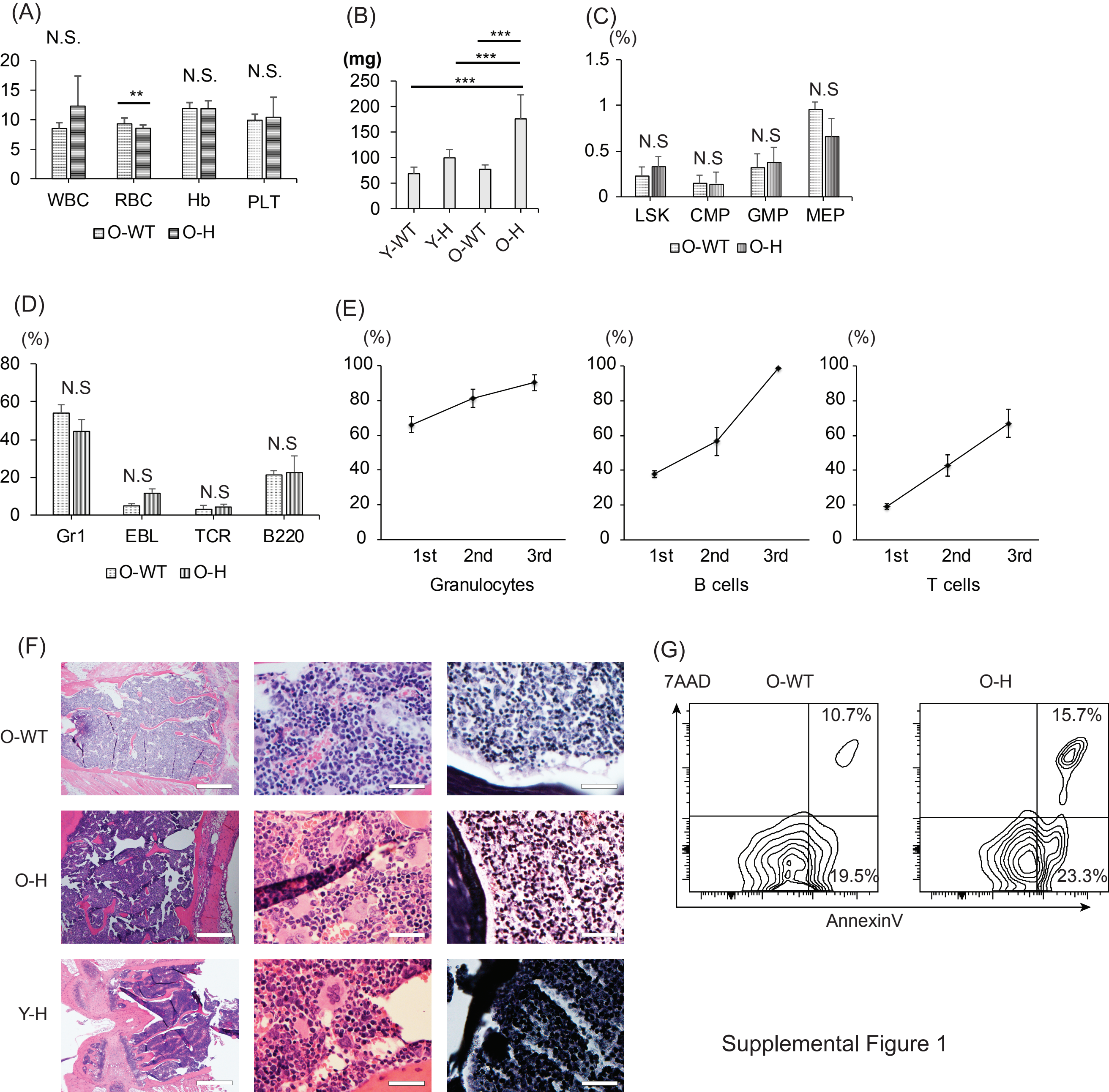

Supplemental Figure 1

**Supplemental Figure 1. ∆*Hmga2* mice develop a mild MDS/MPN-like disease after a long term.**

(A) Complete blood cell count of aged (60-week-old) mice. N = 8 for O-WT and 16 for O-H. **, P < 0.01 by a Student's *t*-test. WBC, white blood cells (× $10^9$/L); RBC (× $10^{12}$/L); Hb, hemoglobin (g/dL); and PLT, platelets (× $10^{11}$/L).

(B) Spleen weight. N = 11 for Y-WT, 9 Y-H, 12 O-WT, and 11 O-H. ***, P < 0.001 (Tukey-HSD test).

(C) The ratio of stem and progenitor cells and (D) differentiated cells in BM-MNCs (N = 3 - 5). The difference was evaluated by a Student's *t*-test.

(E) Competitive serial BMT from O-H mice (Ly5.2$^+$) to lethally irradiated Ly5.1$^+$ mice. The BMT was repeated every 20 weeks. The chimerism of Ly5.2$^+$ cells is shown (N = 3 for each).

(F) BM histology of O-WT, O-H, and Y-H mice. Hematoxylin-eosin (upper; x 40 and middle; x 400) and silver (lower) stains are shown. The scale bars indicate 500 µm for the upper and 50 µm for the middle and lower panels.

(G) Apoptotic CD71$^+$Ter119$^+$ erythroblasts (Annexin V/7AAD staining). Similar results were obtained by three independent experiments.

(A-E) Bars show the means ± SEM.

(A-G) Y-WT, young WT (8-week-old); O-WT, old WT (60-week-old); Y-H, young ∆*Hmga2*; O-H, old ∆*Hmga2*.

**Supplemental Table 1. The primers used for quantitative RT-PCR.**

| Gene | Species | Description | Forward primers | Reverse primers |
|---|---|---|---|---|
| *Actin* | mouse | SYBR green | CATCCGTAAAGACCTCTATGCCAAC | ATGGAGCCACCGATCCACA |
| *Stat3* | mouse | SYBR green | AATGGAAATTGCCCGGATC | AGGCGAGACTCTTCCCACAG |
| *HPRT1* | human | TaqMan | Hs99999909_m1 (LifeTechnologies) | |
| *HMAG2* | human | TaqMan | Hs00171569_m1 (LifeTechnologies) | |
| *U6* | human | TaqMan | Assay ID 001973 (LifeTechnologies) | |
| *MIRlet-7a* | human | TaqMan | Assay ID 000377 (LifeTechnologies) | |
| *MIRlet-7b* | human | TaqMan | Assay ID 002619 (LifeTechnologies) | |
| *MIRlet-7c* | human | TaqMan | Assay ID 000379 (LifeTechnologies) | |

**Supplemental Table 2. Upregulated genes in RNAseq.**

| JE and HJ compared to WT | JE and HJ compared to J |
|---|---|
| 0610040B10Rik | 1700048O20Rik |
| 0610040J01Rik | 1700084E18Rik |
| 1700001G17Rik | 1810041H14Rik |
| 1700001O22Rik | 2310010G23Rik |
| 1700012D01Rik | 2310033P09Rik |
| 1700120K04Rik | 2310075C17Rik |
| 2010111I01Rik | 2610305D13Rik |
| 2310010J17Rik | 2900076A07Rik |
| 2810429I04Rik | 4930512M02Rik |
| 3110070M22Rik | 4930533N22Rik |
| 4930557K07Rik | 5730408K05Rik |
| 4933404O12Rik | 5830444B04Rik |
| 5730408K05Rik | 9030617O03Rik |
| 5730559C18Rik | A330069E16Rik |
| 6030498E09Rik | AA465934 |
| 8430426J06Rik | Ahi1 |
| 9030617O03Rik | Aldh1l1 |
| A930041C12Rik | Aldoc |
| AA465934 | Ankrd13d |
| Abca4 | Ankrd37 |
| Abhd17b | Arhgef4 |
| Ackr1 | Arl5c |
| Acp5 | Asns |
| Actr3b | Aspa |
| Adam9 | BC022687 |
| Adamts17 | Bach2os |
| Agpat5 | Blnk |
| Akr1c12 | Ccdc126 |
| Akr1c19 | Ccl8 |
| Alox5 | Ccr7 |
| Amigo3 | Cd302 |
| Ankrd28 | Cd6 |
| Ap3s1 | Cdk5rap1 |
| Apc2 | Cenpv |
| Apol10b | Chad |
| Arhgdig | Chpf |

Arid5a
Arx
Asns
Ass1
Atp1a3
Atp8b4
B4galnt4
BC021614
Bcas1
Bcat1
Bcl3
Bdh1
Bhlhe40
C2cd4b
C4b
Camk2a
Camk2b
Car12
Card9
Cbr3
Ccdc109b
Ccdc64
Ccl6
Ccne1
Ccng1
Ccnj
Ccr1
Ccr2
Ccr5
Ccrn4l
Cd14
Cd200r3
Cd38
Cdkn1a
Cers6
Chad
Chd7
Chst13
Clca3a1
Cib2
Cnn3
Cox6a2
Cstad
Cx3cr1
Cyp2r1
Cyp51
Degs2
Dhx58
Dpcd
Drc7
Dusp9
E130304I02Rik
Efemp2
Emc8-1190005i06rik
Epb4.1l4aos
Evl
Fam71e1
Fdps
Fxyd1
Gapt
Gcat
Gm13139
Gm13152
Gm15350
Gm15471
Gm20319
Gm38431
Gm38670
Gm5424
Gm5547
Gm7008
Gnat2
Gpc1
Gpr25
Gpx7
H2-Q8
H2-T24
Hebp1

Clec10a
Clnk
Clu
Col18a1
Cox4i2
Csf1r
Csf2rb
Csf2rb2
Csprs
Cst7
Ctsg
Ctsz
Cx3cr1
Cxcl2
Cxcr2
Cyb561
Cyp4f18
Degs2
Dgat2
Dhx40
Dlk1
Dmkn
Dock9
Dtx4
Dusp9
E130012A19Rik
E130304I02Rik
Echdc2
Ecm1
Efemp2
Egln3
Egr4
Ehd3
Emilin2
Endog
Epb4.1l4aos
Esm1
Evpl
F13a1
Hist1h2ai
Hist1h2bl
Hist1h3g
Hist1h4c
Hist2h2ab
Hist3h2ba
Hopx
Hoxaas3
Hsd17b1
Hspa2
Idi1
Ifi27
Ifi27l2a
Ifi44
Ifit1
Ift43
Il17re
Il27
Insig1
Irf7
Isg15
Kcng1
Kifc3
LOC100041034
LOC664787
Ldlr
Lenep
Lhcgr
Lmo1
Lrp11
Mgst2
Mmp11
Mpst
Mt1
Ndn
Noxo1
Npff
Nrg4
Nucb2

Fabp5
Fads2
Fads3
Fam118b
Fam84b
Fcer1a
Ffar2
Fgf3
Fgr
Fhl3
Fkbp11
Flnb
Fndc3b
Frmd6
Fstl3
Fut8
Gadd45b
Gadd45g
Galr3
Gatm
Gbp3
Gbx2
Gca
Gcat
Gdf3
Gdpd5
Gin1
Gja1
Gm10012
Gm14005
Gm15915
Gm16548
Gm16907
Gm19705
Gm38670
Gm5294
Gm7120
Gpr18
Gpr25
Oas1a
Oas1g
Oas2
Oas3
Oscp1
Pcbd1
Pfn4
Prss16
Psme2b
Ptprf
Pydc3
Pygm
Rnd1
Rtp4
Rundc3a
Samd8
Sap25
Sdc3
41161
Slc16a2
Sqle
St6gal1
Tceal3
Tcf7l1
Terc
Tmem119
Tnfrsf14
Tspan17
Tstd3
Ubtd2
Ugt1a6b
Xlr3a
Zbp1
Zfp248

Gpr84
Gramd3
Gzmb
H2afy2
Habp4
Hbb-b2
Hdc
Helq
Hist1h2ah
Hist1h2be
Hist1h3h
Hist1h4h
Hist1h4k
Hist3h2ba
Hist3h2bb-ps
Hmga1-rs1
Homer2
Hopx
Hotairm1
Hoxaas3
Hp
Hpdl
Hpse
Hs6st2
Hsd17b1
Hvcn1
I830077J02Rik
Id1
Ifitm1
Igfbp7
Il18r1
Il1r1
Il1rl1
Il4ra
Irf2bpl
Jmjd7
Kcne3
Klrb1c
Klrb1f

Klrd1
LOC100041034
LOC664787
Lag3
Ldhal6b
Lmo1
Lrg1
Lrp11
Lsr
Ly6c1
Mapk13
Mapkapk2
Matn2
Mboat1
Mcf2l
Mchr1
Mcpt8
Mcts2
Mgl2
Mgst2
Mir17hg
Mmp14
Moap1
Mpzl3
Ms4a2
Ms4a3
Mt1
Mt2
Mtus1
Muc1
Myd88
Myo19
Myo1e
Naf1
Npff
Nr0b2
Nrtn
Nsf
Ntn4

Nupr1
Olfm1
Oosp1
Oosp2
Opn3
Osm
Otulin
Palm
Papss2
Pcdhgc3
Pcsk9
Pdcd1lg2
Pdgfrb
Pdia5
Pdss1
Perp
Pgam2
Pgs1
Phlda1
Pi16
Pim2
Plcd3
Plekhh3
Plod1
Plod2
Plscr1
Plscr2
Pmaip1
Pnp2
Podxl
Ppfibp1
Ppifos
Ppp3cc
Prodh
Prom1
Prps1
Prss16
Prss57
Prtn3

Ptgir
Ptprf
Pvt1
Rab26os
Rab3il1
Rab44
Rbm3os
Rcvrn
Reep6
Rgs10
Rhbdl2
Rhoj
Ribc1
Rpl30
Rps6ka3
Rras2
Rtn4r
Rundc3a
S100a4
S100a5
S100a6
S1pr3
Samd14
Sbno2
Scgb3a1
Selm
Selp
Sema4a
Sema6b
41161
Serpine2
Sgcb
Sipa1l1
Slc16a1
Slc17a7
Slc19a2
Slc22a4
Slc26a10
Slc39a8

Slc4a8
Slc7a8
Slc9a2
SlcUbl4a
Slfn2
Slpi
Smim13
Snhg10
Snx24
Socs2
Sorl1
Sparc
Spint1
Spire1
Spire2
Spsb1
St6gal1
Sulf2
Sult2b1
Syngr1
Syngr4
Tbrg4
Tceal3
Tcf7
Tctex1d4
Tctn1
Tfrc
Tgfbi
Tgm1
Tha1
Tjp1
Tmem132a
Tmem147
Tmem215
Tomm6os
Tpcn1
Trem1
Trim46
Trpv4

| |
|---|
| Tspan17 |
| Tspo |
| Tstd3 |
| Ubxn11 |
| Ugt1a6b |
| Vldlr |
| Wdfy1 |
| Wfs1 |
| Wipi1 |
| Xlr3a |
| Xylb |
| Zbtb16 |
| Zfp106 |
| Znhit1 |

**Supplemental Table 3. Gene sets enriched in phenotype HJ compared to WT.**

| | GS | SIZE | ES | NES | NOM p-val | FDR q-val |
|---|---|---|---|---|---|---|
| 1 | REACTOME_G1_S_TRANSITION | 103 | 0.71 | 2.02 | 0 | 0.002 |
| 2 | REACTOME_SYNTHESIS_OF_DNA | 90 | 0.72 | 2.02 | 0 | 0.001 |
| 3 | REACTOME_ASSEMBLY_OF_THE_PRE_REPLICATIVE_COMPLEX | 63 | 0.74 | 1.99 | 0 | 0.002 |
| 4 | REACTOME_M_G1_TRANSITION | 77 | 0.72 | 1.98 | 0 | 0.001 |
| 5 | REACTOME_ORC1_REMOVAL_FROM_CHROMATIN | 65 | 0.72 | 1.94 | 0 | 0.003 |
| 6 | REACTOME_S_PHASE | 105 | 0.67 | 1.93 | 0 | 0.003 |
| 7 | REACTOME_CELL_CYCLE_CHECKPOINTS | 110 | 0.66 | 1.92 | 0 | 0.004 |
| 8 | REACTOME_CYCLIN_E_ASSOCIATED_EVENTS_DURING_G1_S_TRANSITION_ | 61 | 0.7 | 1.91 | 0 | 0.004 |
| 9 | REACTOME_DESTABILIZATION_OF_MRNA_BY_AUF1_HNRNP_D0 | 50 | 0.73 | 1.9 | 0 | 0.004 |
| 10 | REACTOME_MITOTIC_G1_G1_S_PHASES | 127 | 0.65 | 1.89 | 0 | 0.005 |
| 11 | REACTOME_SCFSKP2_MEDIATED_DEGRADATION_OF_P27_P21 | 52 | 0.73 | 1.88 | 0 | 0.004 |
| 12 | REACTOME_CDT1_ASSOCIATION_WITH_THE_CDC6_ORC_ORIGIN_COMPLEX | 54 | 0.73 | 1.87 | 0 | 0.005 |
| 13 | REACTOME_ASPARAGINE_N_LINKED_GLYCOSYLATION | 78 | 0.68 | 1.83 | 0 | 0.01 |
| 14 | KEGG_AMINOACYL_TRNA_BIOSYNTHESIS | 41 | 0.73 | 1.82 | 0 | 0.012 |
| 15 | REACTOME_TRANSPORT_OF_MATURE_TRANSCRIPT_TO_CYTOPLASM | 52 | 0.7 | 1.82 | 0 | 0.012 |
| 16 | REACTOME_P53_DEPENDENT_G1_DNA_DAMAGE_RESPONSE | 52 | 0.71 | 1.81 | 0 | 0.012 |
| 17 | REACTOME_ACTIVATION_OF_ATR_IN_RESPONSE_TO_REPLICATION_STRESS | 35 | 0.75 | 1.81 | 0.002 | 0.012 |
| 18 | REACTOME_ER_PHAGOSOME_PATHWAY | 53 | 0.71 | 1.81 | 0 | 0.012 |
| 19 | REACTOME_AUTODEGRADATION_OF_THE_E3_UBIQUITIN_LIGASE_COP1 | 46 | 0.71 | 1.8 | 0 | 0.012 |
| 20 | REACTOME_TRNA_AMINOACYLATION | 42 | 0.72 | 1.8 | 0 | 0.012 |
| 21 | REACTOME_ANTIGEN_PROCESSING_CROSS_PRESENTATION | 65 | 0.67 | 1.8 | 0 | 0.012 |
| 22 | REACTOME_P53_INDEPENDENT_G1_S_DNA_DAMAGE_CHECKPOINT | 48 | 0.69 | 1.79 | 0 | 0.012 |
| 23 | REACTOME_CROSS_PRESENTATION_OF_SOLUBLE_EXOGENOUS_ANTIGENS_ENDOSOMES | 45 | 0.7 | 1.79 | 0.002 | 0.013 |
| 24 | REACTOME_TRANSPORT_OF_MATURE_MRNA_DERIVED_FROM_AN_INTRONLESS_TRANSCRIPT | 32 | 0.74 | 1.78 | 0 | 0.014 |
| 25 | REACTOME_VIF_MEDIATED_DEGRADATION_OF_APOBEC3G | 48 | 0.7 | 1.78 | 0.002 | 0.013 |
| 26 | REACTOME_DNA_STRAND_ELONGATION | 30 | 0.76 | 1.78 | 0 | 0.014 |

|  |  |  |  |  |  |  |
|---|---|---|---|---|---|---|
| 27 | REACTOME_MITOCHONDRIAL_PROTEIN_IMPORT | 46 | 0.7 | 1.77 | 0 | 0.017 |
| 28 | BIOCARTA_PROTEASOME_PATHWAY | 28 | 0.76 | 1.77 | 0 | 0.016 |
| 29 | REACTOME_HOST_INTERACTIONS_OF_HIV_FACTORS | 115 | 0.6 | 1.77 | 0 | 0.016 |
| 30 | REACTOME_CDK_MEDIATED_PHOSPHORYLATION_AND_REMOVAL_OF_CDC6 | 46 | 0.71 | 1.76 | 0 | 0.016 |
| 31 | REACTOME_METABOLISM_OF_NUCLEOTIDES | 69 | 0.65 | 1.76 | 0 | 0.016 |
| 32 | REACTOME_SCF_BETA_TRCP_MEDIATED_DEGRADATION_OF_EMI1 | 48 | 0.69 | 1.76 | 0 | 0.016 |
| 33 | KEGG_PROTEASOME | 44 | 0.71 | 1.76 | 0 | 0.016 |
| 34 | KEGG_ANTIGEN_PROCESSING_AND_PRESENTATION | 50 | 0.68 | 1.76 | 0 | 0.015 |
| 35 | REACTOME_METABOLISM_OF_PROTEINS | 397 | 0.54 | 1.76 | 0 | 0.015 |
| 36 | REACTOME_G2_M_CHECKPOINTS | 41 | 0.7 | 1.75 | 0.002 | 0.018 |
| 37 | REACTOME_METABOLISM_OF_NON_CODING_RNA | 47 | 0.7 | 1.75 | 0 | 0.018 |
| 38 | REACTOME_REGULATION_OF_MRNA_STABILITY_BY_PROTEINS_THAT_BIND_AU_RICH_ELEMENTS | 81 | 0.63 | 1.74 | 0 | 0.018 |
| 39 | REACTOME_INTERACTIONS_OF_VPR_WITH_HOST_CELLULAR_PROTEINS | 31 | 0.74 | 1.74 | 0 | 0.018 |
| 40 | REACTOME_DNA_REPLICATION | 184 | 0.57 | 1.74 | 0 | 0.018 |
| 41 | REACTOME_POST_TRANSLATIONAL_PROTEIN_MODIFICATION | 167 | 0.57 | 1.74 | 0 | 0.018 |
| 42 | REACTOME_PURINE_METABOLISM | 32 | 0.74 | 1.73 | 0.002 | 0.018 |
| 43 | REACTOME_REGULATION_OF_GLUCOKINASE_BY_GLUCOKINASE_REGULATORY_PROTEIN | 27 | 0.76 | 1.73 | 0.004 | 0.018 |
| 44 | REACTOME_RESPIRATORY_ELECTRON_TRANSPORT_ATP_SYNTHESIS_BY_CHEMIOSMOTIC_COUPLING_AND_HEAT_PRODUCTION_BY_UNCOUPLING_PROTEINS_ | 77 | 0.65 | 1.73 | 0 | 0.018 |
| 45 | REACTOME_TRANSPORT_OF_RIBONUCLEOPROTEINS_INTO_THE_HOST_NUCLEUS | 27 | 0.75 | 1.73 | 0.002 | 0.019 |
| 46 | REACTOME_GLUCOSE_TRANSPORT | 38 | 0.7 | 1.72 | 0.004 | 0.019 |
| 47 | REACTOME_NEP_NS2_INTERACTS_WITH_THE_CELLULAR_EXPORT_MACHINERY | 27 | 0.76 | 1.72 | 0.004 | 0.019 |
| 48 | REACTOME_REGULATION_OF_ORNITHINE_DECARBOXYLASE_ODC | 48 | 0.67 | 1.71 | 0.004 | 0.022 |
| 49 | REACTOME_EXTENSION_OF_TELOMERES | 27 | 0.73 | 1.71 | 0.008 | 0.024 |
| 50 | KEGG_DNA_REPLICATION | 35 | 0.72 | 1.7 | 0.008 | 0.024 |
| 51 | REACTOME_PROCESSING_OF_CAPPED_INTRON_CONTAINING_PRE_MRNA | 134 | 0.58 | 1.7 | 0 | 0.025 |
| 52 | REACTOME_MRNA_PROCESSING | 151 | 0.57 | 1.7 | 0 | 0.024 |
| 53 | REACTOME_METABOLISM_OF_AMINO_ACIDS_AND_DERIVATIVES | 190 | 0.55 | 1.7 | 0 | 0.024 |
| 54 | REACTOME_E2F_MEDIATED_REGULATION_OF_DNA_REPLICATION | 32 | 0.72 | 1.69 | 0.002 | 0.025 |

|  |  |  |  |  |  |  |
|---|---|---|---|---|---|---|
| 55 | REACTOME_SIGNALING_BY_WNT | 61 | 0.64 | 1.69 | 0 | 0.025 |
| 56 | REACTOME_AUTODEGRADATION_OF_CDH1_BY_CDH1_APC_C | 56 | 0.65 | 1.69 | 0.004 | 0.025 |
| 57 | REACTOME_ACTIVATION_OF_THE_PRE_REPLICATIVE_COMPLEX | 29 | 0.74 | 1.69 | 0.002 | 0.026 |
| 58 | REACTOME_CYTOSOLIC_TRNA_AMINOACYLATION | 24 | 0.77 | 1.69 | 0.002 | 0.025 |
| 59 | KEGG_TERPENOID_BACKBONE_BIOSYNTHESIS | 15 | 0.84 | 1.69 | 0.002 | 0.027 |
| 60 | REACTOME_ACTIVATED_POINT_MUTANTS_OF_FGFR2 | 16 | 0.82 | 1.68 | 0.006 | 0.027 |
| 61 | REACTOME_AMINE_DERIVED_HORMONES | 15 | 0.83 | 1.68 | 0 | 0.027 |
| 62 | REACTOME_RESPIRATORY_ELECTRON_TRANSPORT | 62 | 0.63 | 1.68 | 0.005 | 0.028 |
| 63 | REACTOME_APC_C_CDH1_MEDIATED_DEGRADATION_OF_CDC20_AND_OTHER_APC_C_CDH1_TARGETED_PROTEINS_IN_LATE_MITOSIS_EARLY_G1 | 64 | 0.63 | 1.67 | 0.002 | 0.03 |
| 64 | REACTOME_ACTIVATION_OF_NF_KAPPAB_IN_B_CELLS | 60 | 0.64 | 1.66 | 0.004 | 0.032 |
| 65 | REACTOME_IL_RECEPTOR_SHC_SIGNALING | 27 | 0.73 | 1.66 | 0.011 | 0.033 |
| 66 | REACTOME_RNA_POL_I_PROMOTER_OPENING | 44 | 0.67 | 1.65 | 0.002 | 0.036 |
| 67 | REACTOME_TCA_CYCLE_AND_RESPIRATORY_ELECTRON_TRANSPORT | 112 | 0.57 | 1.65 | 0 | 0.036 |
| 68 | REACTOME_MITOTIC_M_M_G1_PHASES | 164 | 0.55 | 1.65 | 0 | 0.036 |
| 69 | KEGG_CITRATE_CYCLE_TCA_CYCLE | 28 | 0.72 | 1.65 | 0.006 | 0.036 |
| 70 | PID_UPA_UPAR_PATHWAY | 42 | 0.67 | 1.65 | 0.002 | 0.038 |
| 71 | KEGG_GLYCINE_SERINE_AND_THREONINE_METABOLISM | 31 | 0.69 | 1.64 | 0.006 | 0.039 |
| 72 | REACTOME_HIV_INFECTION | 179 | 0.54 | 1.63 | 0 | 0.042 |
| 73 | REACTOME_APC_C_CDC20_MEDIATED_DEGRADATION_OF_MITOTIC_PROTEINS | 65 | 0.61 | 1.63 | 0.002 | 0.043 |
| 74 | REACTOME_CELL_CYCLE_MITOTIC | 300 | 0.51 | 1.63 | 0 | 0.043 |
| 75 | REACTOME_LATE_PHASE_OF_HIV_LIFE_CYCLE | 92 | 0.58 | 1.63 | 0.002 | 0.043 |
| 76 | REACTOME_TELOMERE_MAINTENANCE | 62 | 0.62 | 1.63 | 0.006 | 0.044 |
| 77 | REACTOME_CELL_CYCLE | 377 | 0.49 | 1.63 | 0 | 0.043 |
| 78 | KEGG_SPLICEOSOME | 121 | 0.56 | 1.61 | 0 | 0.05 |
| 79 | PID_E2F_PATHWAY | 69 | 0.6 | 1.61 | 0 | 0.051 |
| 80 | REACTOME_STEROID_HORMONES | 27 | 0.69 | 1.61 | 0.006 | 0.051 |
| 81 | REACTOME_REGULATION_OF_MITOTIC_CELL_CYCLE | 76 | 0.59 | 1.61 | 0.002 | 0.052 |
| 82 | REACTOME_TRANSPORT_TO_THE_GOLGI_AND_SUBSEQUENT_MODIFICATION | 32 | 0.68 | 1.6 | 0.008 | 0.056 |

| | | | | | | |
|---|---|---|---|---|---|---|
| 83 | KEGG_ARGININE_AND_PROLINE_METABOLISM | 49 | 0.63 | 1.6 | 0.012 | 0.056 |
| 84 | REACTOME_ABC_FAMILY_PROTEINS_MEDIATED_TRANSPORT | 29 | 0.7 | 1.59 | 0.013 | 0.061 |
| 85 | REACTOME_AMYLOIDS | 61 | 0.61 | 1.58 | 0.008 | 0.063 |
| 86 | REACTOME_G1_S_SPECIFIC_TRANSCRIPTION | 17 | 0.76 | 1.58 | 0.015 | 0.068 |
| 87 | REACTOME_CITRIC_ACID_CYCLE_TCA_CYCLE | 18 | 0.75 | 1.56 | 0.023 | 0.077 |
| 88 | REACTOME_METABOLISM_OF_STEROID_HORMONES_AND_VITAMINS_A_AND_D | 32 | 0.67 | 1.56 | 0.019 | 0.076 |
| 89 | REACTOME_TRANSCRIPTION | 175 | 0.52 | 1.56 | 0 | 0.076 |
| 90 | REACTOME_LAGGING_STRAND_SYNTHESIS | 19 | 0.74 | 1.56 | 0.02 | 0.079 |
| 91 | KEGG_SYSTEMIC_LUPUS_ERYTHEMATOSUS | 92 | 0.55 | 1.56 | 0.004 | 0.079 |
| 92 | KEGG_SELENOAMINO_ACID_METABOLISM | 25 | 0.69 | 1.55 | 0.012 | 0.081 |
| 93 | BIOCARTA_MCM_PATHWAY | 17 | 0.77 | 1.55 | 0.015 | 0.082 |
| 94 | REACTOME_SYNTHESIS_SECRETION_AND_INACTIVATION_OF_GLP1 | 17 | 0.75 | 1.55 | 0.033 | 0.086 |
| 95 | KEGG_MISMATCH_REPAIR | 22 | 0.71 | 1.54 | 0.023 | 0.086 |
| 96 | REACTOME_INCRETIN_SYNTHESIS_SECRETION_AND_INACTIVATION | 20 | 0.72 | 1.54 | 0.019 | 0.087 |
| 97 | KEGG_N_GLYCAN_BIOSYNTHESIS | 44 | 0.62 | 1.54 | 0.004 | 0.089 |
| 98 | REACTOME_MRNA_SPLICING | 105 | 0.53 | 1.54 | 0.004 | 0.09 |
| 99 | PID_ATR_PATHWAY | 39 | 0.63 | 1.53 | 0.021 | 0.091 |
| 100 | REACTOME_DIABETES_PATHWAYS | 120 | 0.52 | 1.53 | 0.002 | 0.094 |
| 101 | REACTOME_PACKAGING_OF_TELOMERE_ENDS | 35 | 0.63 | 1.53 | 0.024 | 0.097 |
| 102 | KEGG_OXIDATIVE_PHOSPHORYLATION | 112 | 0.52 | 1.52 | 0.002 | 0.102 |
| 103 | REACTOME_HIV_LIFE_CYCLE | 105 | 0.53 | 1.52 | 0.011 | 0.103 |
| 104 | REACTOME_UNFOLDED_PROTEIN_RESPONSE | 73 | 0.56 | 1.51 | 0.015 | 0.107 |
| 105 | REACTOME_SLC_MEDIATED_TRANSMEMBRANE_TRANSPORT | 233 | 0.48 | 1.51 | 0 | 0.109 |
| 106 | REACTOME_PROCESSIVE_SYNTHESIS_ON_THE_LAGGING_STRAND | 15 | 0.74 | 1.5 | 0.03 | 0.115 |
| 107 | REACTOME_MEIOTIC_RECOMBINATION | 66 | 0.57 | 1.5 | 0.009 | 0.114 |
| 108 | REACTOME_N_GLYCAN_ANTENNAE_ELONGATION_IN_THE_MEDIAL_TRANS_GOLGI | 18 | 0.73 | 1.5 | 0.05 | 0.116 |
| 109 | REACTOME_METABOLISM_OF_RNA | 249 | 0.46 | 1.5 | 0 | 0.115 |
| 110 | REACTOME_CLEAVAGE_OF_GROWING_TRANSCRIPT_IN_THE_TERMINATION_REGION_ | 41 | 0.61 | 1.49 | 0.029 | 0.123 |
| 111 | NABA_ECM_REGULATORS | 212 | 0.48 | 1.49 | 0 | 0.128 |

| | | | | | | |
|---|---|---|---|---|---|---|
| 112 | KEGG_LYSOSOME | 117 | 0.51 | 1.48 | 0.012 | 0.13 |
| 113 | REACTOME_MRNA_SPLICING_MINOR_PATHWAY | 41 | 0.6 | 1.48 | 0.021 | 0.136 |
| 114 | KEGG_ABC_TRANSPORTERS | 40 | 0.6 | 1.47 | 0.026 | 0.137 |
| 115 | REACTOME_SYNTHESIS_AND_INTERCONVERSION_OF_NUCLEOTIDE_DI_AND_TRIPHOSPHATES | 17 | 0.73 | 1.47 | 0.048 | 0.139 |
| 116 | C | 15 | 0.75 | 1.47 | 0.044 | 0.139 |
| 117 | REACTOME_MRNA_3_END_PROCESSING | 33 | 0.62 | 1.47 | 0.026 | 0.138 |
| 118 | REACTOME_RNA_POL_I_RNA_POL_III_AND_MITOCHONDRIAL_TRANSCRIPTION | 99 | 0.51 | 1.46 | 0.021 | 0.151 |
| 119 | PID_MYC_ACTIV_PATHWAY | 77 | 0.53 | 1.46 | 0.006 | 0.15 |
| 120 | REACTOME_RNA_POL_II_TRANSCRIPTION | 91 | 0.52 | 1.46 | 0.008 | 0.15 |
| 121 | REACTOME_INFLUENZA_LIFE_CYCLE | 131 | 0.49 | 1.45 | 0.007 | 0.153 |
| 122 | KEGG_OTHER_GLYCAN_DEGRADATION | 16 | 0.71 | 1.45 | 0.036 | 0.155 |
| 123 | REACTOME_TRANSLATION | 138 | 0.49 | 1.45 | 0.004 | 0.156 |
| 124 | REACTOME_REGULATION_OF_apoptosis | 56 | 0.55 | 1.45 | 0.025 | 0.157 |
| 125 | REACTOME_ACTIVATION_OF_chaperone_GENES_BY_XBP1S | 42 | 0.58 | 1.45 | 0.023 | 0.157 |
| 126 | REACTOME_RNA_POL_I_TRANSCRIPTION | 68 | 0.54 | 1.44 | 0.025 | 0.158 |
| 127 | KEGG_apoptosis | 79 | 0.52 | 1.44 | 0.011 | 0.159 |
| 128 | KEGG_GALACTOSE_METABOLISM | 25 | 0.64 | 1.44 | 0.045 | 0.158 |
| 129 | REACTOME_TRANSMEMBRANE_TRANSPORT_OF_SMALL_MOLECULES | 391 | 0.43 | 1.44 | 0 | 0.164 |
| 130 | REACTOME_TAK1_ACTIVATES_NFKB_BY_PHOSPHORYLATION_AND_ACTIVATION_OF_IKKS_COMPLEX | 22 | 0.66 | 1.43 | 0.04 | 0.164 |
| 131 | REACTOME_TRANSCRIPTION_COUPLED_NER_TC_NER | 43 | 0.58 | 1.43 | 0.034 | 0.166 |
| 132 | PID_FANCONI_PATHWAY | 45 | 0.57 | 1.42 | 0.038 | 0.175 |
| 133 | REACTOME_DEPOSITION_OF_NEW_CENPA_CONTAINING_NUCLEOSOMES_AT_THE_CENTROMERE | 50 | 0.55 | 1.42 | 0.041 | 0.176 |
| 134 | REACTOME_EXTRACELLULAR_MATRIX_ORGANIZATION | 79 | 0.51 | 1.42 | 0.019 | 0.175 |
| 135 | REACTOME_BIOSYNTHESIS_OF_THE_N_GLYCAN_PRECURSOR_DOLICHOL_LIPID_LINKED_OLIGOSACCHARIDE_LLO_AND_TRANSFER_TO_A_NASCENT_PROTEIN | 27 | 0.62 | 1.41 | 0.043 | 0.18 |
| 136 | KEGG_HISTIDINE_METABOLISM | 27 | 0.62 | 1.41 | 0.049 | 0.179 |
| 137 | KEGG_PYRIMIDINE_METABOLISM | 95 | 0.5 | 1.41 | 0.017 | 0.18 |
| 138 | REACTOME_CHROMOSOME_MAINTENANCE | 104 | 0.49 | 1.4 | 0.002 | 0.186 |
| 139 | REACTOME_FORMATION_OF_FIBRIN_CLOT_CLOTTING_CASCADE | 31 | 0.59 | 1.4 | 0.049 | 0.189 |

| | | | | | | |
|---|---|---|---|---|---|---|
| 140 | REACTOME_TRANS_GOLGI_NETWORK_VESICLE_BUDDING | 54 | 0.53 | 1.38 | 0.042 | 0.216 |
| 141 | REACTOME_G_ALPHA_I_SIGNALLING_EVENTS | 176 | 0.45 | 1.37 | 0.014 | 0.226 |
| 142 | REACTOME_MEMBRANE_TRAFFICKING | 117 | 0.47 | 1.36 | 0.023 | 0.235 |
| 143 | KEGG_PARKINSONS_DISEASE | 109 | 0.48 | 1.36 | 0.029 | 0.237 |
| 144 | REACTOME_CLASS_A1_RHODOPSIN_LIKE_RECEPTORS | 263 | 0.42 | 1.35 | 0.002 | 0.247 |

**Supplemental Table 4. Gene sets enriched in phenotype HJ compared to J (LSK).**

| | GS | SIZE | ES | NES | NOM p-val | FDR q-val |
|---|---|---|---|---|---|---|
| 1 | REACTOME_SYNTHESIS_OF_DNA | 90 | 0.72 | 2.03 | 0 | 0.005 |
| 2 | REACTOME_M_G1_TRANSITION | 77 | 0.72 | 2 | 0 | 0.006 |
| 3 | REACTOME_S_PHASE | 105 | 0.69 | 2 | 0 | 0.004 |
| 4 | REACTOME_G1_S_TRANSITION | 103 | 0.68 | 1.98 | 0 | 0.006 |
| 5 | REACTOME_ASSEMBLY_OF_THE_PRE_REPLICATIVE_COMPLEX | 63 | 0.74 | 1.97 | 0 | 0.005 |
| 6 | REACTOME_CHOLESTEROL_BIOSYNTHESIS | 20 | 0.89 | 1.96 | 0 | 0.005 |
| 7 | REACTOME_ORC1_REMOVAL_FROM_CHROMATIN | 65 | 0.72 | 1.94 | 0 | 0.006 |
| 8 | REACTOME_CDT1_ASSOCIATION_WITH_THE_CDC6_ORC_ORIGIN_COMPLEX | 54 | 0.73 | 1.92 | 0 | 0.009 |
| 9 | REACTOME_RNA_POL_I_PROMOTER_OPENING | 44 | 0.75 | 1.9 | 0 | 0.01 |
| 10 | REACTOME_MITOTIC_G1_G1_S_PHASES | 127 | 0.63 | 1.9 | 0 | 0.009 |
| 11 | REACTOME_DESTABILIZATION_OF_MRNA_BY_AUF1_HNRNP_D0 | 50 | 0.72 | 1.88 | 0 | 0.011 |
| 12 | REACTOME_DNA_STRAND_ELONGATION | 30 | 0.79 | 1.86 | 0 | 0.013 |
| 13 | BIOCARTA_MCM_PATHWAY | 17 | 0.87 | 1.85 | 0 | 0.014 |
| 14 | REACTOME_CYCLIN_E_ASSOCIATED_EVENTS_DURING_G1_S_TRANSITION_ | 61 | 0.68 | 1.83 | 0 | 0.02 |
| 15 | REACTOME_ANTIGEN_PROCESSING_CROSS_PRESENTATION | 65 | 0.68 | 1.82 | 0 | 0.02 |
| 16 | REACTOME_CDK_MEDIATED_PHOSPHORYLATION_AND_REMOVAL_OF_CDC6 | 46 | 0.71 | 1.82 | 0 | 0.02 |
| 17 | REACTOME_VIF_MEDIATED_DEGRADATION_OF_APOBEC3G | 48 | 0.69 | 1.81 | 0.002 | 0.02 |
| 18 | REACTOME_ACTIVATION_OF_THE_PRE_REPLICATIVE_COMPLEX | 29 | 0.78 | 1.81 | 0 | 0.019 |
| 19 | KEGG_DNA_REPLICATION | 35 | 0.75 | 1.81 | 0 | 0.019 |
| 20 | KEGG_ANTIGEN_PROCESSING_AND_PRESENTATION | 50 | 0.7 | 1.81 | 0 | 0.018 |
| 21 | REACTOME_AUTODEGRADATION_OF_THE_E3_UBIQUITIN_LIGASE_COP1 | 46 | 0.7 | 1.81 | 0.004 | 0.017 |
| 22 | REACTOME_P53_DEPENDENT_G1_DNA_DAMAGE_RESPONSE | 52 | 0.7 | 1.8 | 0.004 | 0.019 |
| 23 | REACTOME_ER_PHAGOSOME_PATHWAY | 53 | 0.7 | 1.8 | 0 | 0.019 |

| | | | | | | |
|---|---|---|---|---|---|---|
| 24 | REACTOME_TRANSLATION | 138 | 0.59 | 1.79 | 0 | 0.019 |
| 25 | REACTOME_INFLUENZA_LIFE_CYCLE | 131 | 0.59 | 1.78 | 0 | 0.022 |
| 26 | REACTOME_ACTIVATION_OF_ATR_IN_RESPONSE_TO_REPLICATION_STRESS | 35 | 0.73 | 1.76 | 0.002 | 0.024 |
| 27 | REACTOME_MEIOTIC_RECOMBINATION | 66 | 0.66 | 1.76 | 0 | 0.025 |
| 28 | REACTOME_SCFSKP2_MEDIATED_DEGRADATION_OF_P27_P21 | 52 | 0.68 | 1.76 | 0.002 | 0.025 |
| 29 | REACTOME_REGULATION_OF_apoptosis | 56 | 0.66 | 1.75 | 0.002 | 0.025 |
| 30 | REACTOME_CROSS_PRESENTATION_OF_SOLUBLE_EXOGENOUS_ANTIGENS_E | 45 | 0.68 | 1.75 | 0 | 0.025 |
| 31 | KEGG_PROTEASOME | 44 | 0.7 | 1.75 | 0.002 | 0.026 |
| 32 | KEGG_SYSTEMIC_LUPUS_ERYTHEMATOSUS | 92 | 0.62 | 1.74 | 0 | 0.027 |
| 33 | BIOCARTA_PROTEASOME_PATHWAY | 28 | 0.72 | 1.74 | 0.004 | 0.027 |
| 34 | KEGG_SPLICEOSOME | 121 | 0.59 | 1.73 | 0 | 0.028 |
| 35 | REACTOME_P53_INDEPENDENT_G1_S_DNA_DAMAGE_CHECKPOINT | 48 | 0.68 | 1.73 | 0 | 0.027 |
| 36 | REACTOME_SRP_DEPENDENT_COTRANSLATIONAL_PROTEIN_TARGETING_TO_ | 103 | 0.6 | 1.73 | 0 | 0.027 |
| 37 | REACTOME_STEROID_HORMONES | 27 | 0.73 | 1.73 | 0 | 0.027 |
| 38 | REACTOME_TELOMERE_MAINTENANCE | 62 | 0.64 | 1.72 | 0.002 | 0.028 |
| 39 | REACTOME_METABOLISM_OF_STEROID_HORMONES_AND_VITAMINS_A_AND_D | 32 | 0.73 | 1.72 | 0.002 | 0.028 |
| 40 | REACTOME_SCF_BETA_TRCP_MEDIATED_DEGRADATION_OF_EMI1 | 48 | 0.67 | 1.72 | 0 | 0.027 |
| 41 | REACTOME_NUCLEOTIDE_EXCISION_REPAIR | 48 | 0.67 | 1.72 | 0.004 | 0.027 |
| 42 | REACTOME_CELL_CYCLE_CHECKPOINTS | 110 | 0.58 | 1.71 | 0 | 0.028 |
| 43 | REACTOME_TRANSCRIPTION_COUPLED_NER_TC_NER | 43 | 0.67 | 1.71 | 0.002 | 0.028 |
| 44 | REACTOME_AMYLOIDS | 61 | 0.64 | 1.7 | 0.005 | 0.031 |
| 45 | KEGG_PORPHYRIN_AND_CHLOROPHYLL_METABOLISM | 29 | 0.73 | 1.7 | 0.002 | 0.031 |
| 46 | REACTOME_GLOBAL_GENOMIC_NER_GG_NER | 32 | 0.72 | 1.69 | 0.011 | 0.031 |
| 47 | REACTOME_EXTENSION_OF_TELOMERES | 27 | 0.74 | 1.69 | 0.002 | 0.031 |
| 48 | REACTOME_PACKAGING_OF_TELOMERE_ENDS | 35 | 0.7 | 1.68 | 0.002 | 0.035 |
| 49 | REACTOME_METABOLISM_OF_PROTEINS | 397 | 0.5 | 1.67 | 0 | 0.037 |

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|---|---|---|---|---|---|---|
| 50 | REACTOME_3_UTR_MEDIATED_TRANSLATIONAL_REGULATION | 99 | 0.58 | 1.67 | 0.004 | 0.037 |
| 51 | REACTOME_TRANSCRIPTION | 175 | 0.53 | 1.66 | 0 | 0.038 |
| 52 | REACTOME_VOLTAGE_GATED_POTASSIUM_CHANNELS | 43 | 0.65 | 1.66 | 0.004 | 0.04 |
| 53 | REACTOME_RNA_POL_I_TRANSCRIPTION | 68 | 0.61 | 1.65 | 0.005 | 0.043 |
| 54 | REACTOME_MITOCHONDRIAL_PROTEIN_IMPORT | 46 | 0.64 | 1.64 | 0.004 | 0.048 |
| 55 | REACTOME_G1_S_SPECIFIC_TRANSCRIPTION | 17 | 0.78 | 1.63 | 0.002 | 0.05 |
| 56 | REACTOME_METABOLISM_OF_NON_CODING_RNA | 47 | 0.63 | 1.63 | 0.005 | 0.05 |
| 57 | REACTOME_LATE_PHASE_OF_HIV_LIFE_CYCLE | 92 | 0.57 | 1.62 | 0.002 | 0.051 |
| 58 | REACTOME_REGULATION_OF_ORNITHINE_DECARBOXYLASE_ODC | 48 | 0.63 | 1.62 | 0.011 | 0.052 |
| 59 | KEGG_NUCLEOTIDE_EXCISION_REPAIR | 43 | 0.63 | 1.62 | 0.011 | 0.052 |
| 60 | REACTOME_INFLUENZA_VIRAL_RNA_TRANSCRIPTION_AND_REPLICATION | 97 | 0.56 | 1.62 | 0.005 | 0.052 |
| 61 | REACTOME_FANCONI_ANEMIA_PATHWAY | 19 | 0.76 | 1.62 | 0.015 | 0.051 |
| 62 | REACTOME_G2_M_CHECKPOINTS | 41 | 0.65 | 1.62 | 0.002 | 0.051 |
| 63 | KEGG_STEROID_BIOSYNTHESIS | 15 | 0.79 | 1.61 | 0.011 | 0.055 |
| 64 | REACTOME_PROCESSING_OF_CAPPED_INTRON_CONTAINING_PRE_MRNA | 134 | 0.53 | 1.61 | 0 | 0.055 |
| 65 | REACTOME_HOST_INTERACTIONS_OF_HIV_FACTORS | 115 | 0.55 | 1.6 | 0.002 | 0.055 |
| 66 | REACTOME_METABOLISM_OF_RNA | 249 | 0.5 | 1.6 | 0 | 0.056 |
| 67 | REACTOME_HIV_INFECTION | 179 | 0.52 | 1.6 | 0 | 0.056 |
| 68 | REACTOME_E2F_MEDIATED_REGULATION_OF_DNA_REPLICATION | 32 | 0.66 | 1.59 | 0.017 | 0.061 |
| 69 | PID_HIF1A_PATHWAY | 18 | 0.75 | 1.59 | 0.026 | 0.061 |
| 70 | REACTOME_DEPOSITION_OF_NEW_CENPA_CONTAINING_NUCLEOSOMES_AT_ | 50 | 0.61 | 1.59 | 0.006 | 0.06 |
| 71 | REACTOME_AUTODEGRADATION_OF_CDH1_BY_CDH1_APC_C | 56 | 0.59 | 1.58 | 0.009 | 0.061 |
| 72 | REACTOME_RNA_POL_I_RNA_POL_III_AND_MITOCHONDRIAL_TRANSCRIPTION | 99 | 0.55 | 1.58 | 0 | 0.061 |
| 73 | REACTOME_MRNA_PROCESSING | 151 | 0.52 | 1.58 | 0 | 0.063 |
| 74 | KEGG_TERPENOID_BACKBONE_BIOSYNTHESIS | 15 | 0.79 | 1.58 | 0.015 | 0.064 |
| 75 | REACTOME_TRNA_AMINOACYLATION | 42 | 0.62 | 1.57 | 0.011 | 0.066 |

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|---|---|---|---|---|---|---|
| 76 | REACTOME_LAGGING_STRAND_SYNTHESIS | 19 | 0.74 | 1.56 | 0.024 | 0.069 |
| 77 | REACTOME_DNA_REPAIR | 101 | 0.55 | 1.56 | 0.005 | 0.069 |
| 78 | KEGG_RIBOSOME | 84 | 0.55 | 1.56 | 0.004 | 0.071 |
| 79 | REACTOME_ASPARAGINE_N_LINKED_GLYCOSYLATION | 78 | 0.57 | 1.55 | 0.016 | 0.074 |
| 80 | REACTOME_REGULATION_OF_MRNA_STABILITY_BY_PROTEINS_THAT_BIND_AU | 81 | 0.55 | 1.54 | 0.007 | 0.082 |
| 81 | REACTOME_PEPTIDE_CHAIN_ELONGATION | 81 | 0.55 | 1.54 | 0.011 | 0.081 |
| 82 | KEGG_MISMATCH_REPAIR | 22 | 0.71 | 1.54 | 0.024 | 0.083 |
| 83 | REACTOME_REGULATION_OF_GLUCOKINASE_BY_GLUCOKINASE_REGULATOR | 27 | 0.66 | 1.53 | 0.016 | 0.087 |
| 84 | REACTOME_NEP_NS2_INTERACTS_WITH_THE_CELLULAR_EXPORT_MACHINER | 27 | 0.66 | 1.53 | 0.02 | 0.089 |
| 85 | REACTOME_TRANSPORT_OF_MATURE_TRANSCRIPT_TO_CYTOPLASM | 52 | 0.58 | 1.52 | 0.021 | 0.089 |
| 86 | REACTOME_CYTOSOLIC_TRNA_AMINOACYLATION | 24 | 0.68 | 1.52 | 0.031 | 0.09 |
| 87 | KEGG_TRYPTOPHAN_METABOLISM | 39 | 0.61 | 1.52 | 0.019 | 0.09 |
| 88 | REACTOME_RNA_POL_II_TRANSCRIPTION | 91 | 0.53 | 1.52 | 0.005 | 0.093 |
| 89 | REACTOME_ACTIVATION_OF_NF_KAPPAB_IN_B_CELLS | 60 | 0.56 | 1.51 | 0.013 | 0.093 |
| 90 | KEGG_GLYCINE_SERINE_AND_THREONINE_METABOLISM | 31 | 0.65 | 1.51 | 0.018 | 0.095 |
| 91 | REACTOME_MRNA_SPLICING_MINOR_PATHWAY | 41 | 0.61 | 1.5 | 0.024 | 0.099 |
| 92 | REACTOME_PROCESSIVE_SYNTHESIS_ON_THE_LAGGING_STRAND | 15 | 0.74 | 1.5 | 0.042 | 0.099 |
| 93 | REACTOME_METABOLISM_OF_MRNA | 205 | 0.48 | 1.5 | 0 | 0.104 |
| 94 | REACTOME_MRNA_SPLICING | 105 | 0.51 | 1.49 | 0.009 | 0.107 |
| 95 | REACTOME_OXYGEN_DEPENDENT_PROLINE_HYDROXYLATION_OF_HYPOXIA_ | 15 | 0.74 | 1.49 | 0.036 | 0.107 |
| 96 | REACTOME_HIV_LIFE_CYCLE | 105 | 0.51 | 1.48 | 0.007 | 0.112 |
| 97 | REACTOME_MEIOSIS | 93 | 0.53 | 1.48 | 0.015 | 0.112 |
| 98 | REACTOME_SIGNALING_BY_WNT | 61 | 0.56 | 1.48 | 0.03 | 0.114 |
| 99 | KEGG_SELENOAMINO_ACID_METABOLISM | 25 | 0.64 | 1.47 | 0.032 | 0.12 |
| 100 | REACTOME_FORMATION_OF_RNA_POL_II_ELONGATION_COMPLEX_ | 35 | 0.59 | 1.46 | 0.026 | 0.129 |
| 101 | REACTOME_TRANSPORT_OF_RIBONUCLEOPROTEINS_INTO_THE_HOST_NUCL | 27 | 0.61 | 1.46 | 0.045 | 0.132 |

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|---|---|---|---|---|---|---|
| 102 | KEGG_CITRATE_CYCLE_TCA_CYCLE | 28 | 0.63 | 1.45 | 0.041 | 0.134 |
| 103 | REACTOME_TRANSPORT_OF_MATURE_MRNA_DERIVED_FROM_AN_INTRONLE | 32 | 0.61 | 1.45 | 0.041 | 0.135 |
| 104 | KEGG_HISTIDINE_METABOLISM | 27 | 0.62 | 1.45 | 0.047 | 0.137 |
| 105 | KEGG_AMINOACYL_TRNA_BIOSYNTHESIS | 41 | 0.57 | 1.44 | 0.029 | 0.143 |
| 106 | REACTOME_METABOLISM_OF_VITAMINS_AND_COFACTORS | 50 | 0.55 | 1.44 | 0.03 | 0.143 |
| 107 | REACTOME_POTASSIUM_CHANNELS | 97 | 0.5 | 1.43 | 0.007 | 0.149 |
| 108 | REACTOME_METABOLISM_OF_AMINO_ACIDS_AND_DERIVATIVES | 190 | 0.45 | 1.41 | 0.01 | 0.163 |
| 109 | REACTOME_MEIOTIC_SYNAPSIS | 58 | 0.53 | 1.41 | 0.024 | 0.172 |
| 110 | KEGG_N_GLYCAN_BIOSYNTHESIS | 44 | 0.55 | 1.4 | 0.044 | 0.175 |
| 111 | KEGG_GLUTATHIONE_METABOLISM | 48 | 0.55 | 1.4 | 0.037 | 0.174 |
| 112 | REACTOME_NONSENSE_MEDIATED_DECAY_ENHANCED_BY_THE_EXON_JUNC | 102 | 0.48 | 1.4 | 0.022 | 0.178 |
| 113 | REACTOME_CHEMOKINE_RECEPTORS_BIND_CHEMOKINES | 44 | 0.54 | 1.39 | 0.042 | 0.181 |
| 114 | REACTOME_APC_C_CDH1_MEDIATED_DEGRADATION_OF_CDC20_AND_OTHER | 64 | 0.51 | 1.39 | 0.04 | 0.179 |
| 115 | KEGG_TYROSINE_METABOLISM | 37 | 0.57 | 1.38 | 0.049 | 0.196 |
| 116 | REACTOME_DNA_REPLICATION | 184 | 0.44 | 1.36 | 0.015 | 0.208 |
| 117 | REACTOME_RNA_POL_II_PRE_TRANSCRIPTION_EVENTS | 50 | 0.53 | 1.36 | 0.043 | 0.205 |
| 118 | REACTOME_CHROMOSOME_MAINTENANCE | 104 | 0.46 | 1.35 | 0.041 | 0.211 |
| 119 | REACTOME_POST_TRANSLATIONAL_PROTEIN_MODIFICATION | 167 | 0.44 | 1.34 | 0.024 | 0.22 |

**Supplemental Table 5. Gene sets enriched in phenotype HJ compared to WT (erythroblasts).**

| | GS | SIZE | ES | NES | NOM p-val | FDR q-val |
|---|---|---|---|---|---|---|
| 1 | REACTOME_PEPTIDE_CHAIN_ELONGATION | 76 | 0.54 | 2.27 | 0.000 | 0.000 |
| 2 | KEGG_RIBOSOME | 76 | 0.54 | 2.26 | 0.000 | 0.000 |
| 3 | REACTOME_3_UTR_MEDIATED_TRANSLATIONAL_REGULATION | 82 | 0.53 | 2.22 | 0.000 | 0.001 |
| 4 | REACTOME_FORMATION_OF_THE_TERNARY_COMPLEX_AND_SUBSEQUENTLY_THE_43S_COMPLEX | 34 | 0.62 | 2.22 | 0.000 | 0.001 |
| 5 | REACTOME_ACTIVATION_OF_THE_MRNA_UPON_BINDING_OF_THE_CAP_BINDING_COMPLEX_AND_EIFS_AND_SUBSEQUENT_BINDING_TO_43S | 40 | 0.59 | 2.16 | 0.000 | 0.001 |
| 6 | REACTOME_SRP_DEPENDENT_COTRANSLATIONAL_PROTEIN_TARGETING_TO_MEMBRANE | 87 | 0.50 | 2.14 | 0.000 | 0.001 |
| 7 | REACTOME_TRANSLATION | 103 | 0.48 | 2.12 | 0.000 | 0.001 |
| 8 | REACTOME_INTERFERON_SIGNALING | 52 | 0.53 | 2.09 | 0.000 | 0.002 |
| 9 | REACTOME_NONSENSE_MEDIATED_DECAY_ENHANCED_BY_THE_EXON_JUNCTION_COMPLEX | 88 | 0.49 | 2.07 | 0.000 | 0.002 |
| 10 | REACTOME_INTERFERON_ALPHA_BETA_SIGNALING | 24 | 0.63 | 2.04 | 0.000 | 0.003 |
| 11 | REACTOME_INFLUENZA_LIFE_CYCLE | 93 | 0.46 | 1.99 | 0.000 | 0.004 |
| 12 | REACTOME_INFLUENZA_VIRAL_RNA_TRANSCRIPTION_AND_REPLICATION | 81 | 0.47 | 1.98 | 0.001 | 0.004 |
| 13 | REACTOME_DIABETES_PATHWAYS | 36 | 0.55 | 1.97 | 0.000 | 0.005 |
| 14 | REACTOME_METABOLISM_OF_RNA | 162 | 0.41 | 1.94 | 0.000 | 0.007 |
| 15 | REACTOME_METABOLISM_OF_MRNA | 149 | 0.42 | 1.94 | 0.000 | 0.007 |
| 16 | REACTOME_INTERFERON_GAMMA_SIGNALING | 19 | 0.60 | 1.86 | 0.003 | 0.016 |
| 17 | KEGG_RIG_I_LIKE_RECEPTOR_SIGNALING_PATHWAY | 16 | 0.63 | 1.83 | 0.002 | 0.021 |
| 18 | KEGG_P53_SIGNALING_PATHWAY | 23 | 0.57 | 1.83 | 0.003 | 0.020 |
| 19 | REACTOME_UNFOLDED_PROTEIN_RESPONSE | 27 | 0.55 | 1.80 | 0.005 | 0.028 |

| | | | | | | |
|---|---|---|---|---|---|---|
| 20 | REACTOME_RIG_I_MDA5_MEDIATED_INDUCTION_OF_IFN_ALPHA_BETA_PATHWAYS | 25 | 0.53 | 1.74 | 0.005 | 0.046 |
| 21 | KEGG_PROTEASOME | 41 | 0.47 | 1.72 | 0.003 | 0.053 |
| 22 | REACTOME_P53_DEPENDENT_G1_DNA_DAMAGE_RESPONSE | 47 | 0.46 | 1.72 | 0.003 | 0.054 |
| 23 | REACTOME_ANTIVIRAL_MECHANISM_BY_IFN_STIMULATED_GENES | 24 | 0.52 | 1.71 | 0.002 | 0.056 |
| 24 | REACTOME_DOWNSTREAM_SIGNALING_EVENTS_OF_B_CELL_RECEPTOR_BCR | 64 | 0.42 | 1.69 | 0.003 | 0.065 |
| 25 | REACTOME_ORC1_REMOVAL_FROM_CHROMATIN | 51 | 0.44 | 1.68 | 0.005 | 0.068 |
| 26 | BIOCARTA_PROTEASOME_PATHWAY | 26 | 0.50 | 1.66 | 0.013 | 0.076 |
| 27 | REACTOME_AUTODEGRADATION_OF_CDH1_BY_CDH1_APC_C | 47 | 0.44 | 1.66 | 0.003 | 0.079 |
| 28 | REACTOME_CDT1_ASSOCIATION_WITH_THE_CDC6_ORC_ORIGIN_COMPLEX | 43 | 0.44 | 1.65 | 0.011 | 0.084 |
| 29 | REACTOME_DESTABILIZATION_OF_MRNA_BY_AUF1_HNRNP_D0 | 46 | 0.43 | 1.64 | 0.011 | 0.085 |
| 30 | REACTOME_CDK_MEDIATED_PHOSPHORYLATION_AND_REMOVAL_OF_CDC6 | 43 | 0.44 | 1.64 | 0.009 | 0.084 |
| 31 | REACTOME_ACTIVATION_OF_NF_KAPPAB_IN_B_CELLS | 50 | 0.42 | 1.62 | 0.008 | 0.093 |
| 32 | REACTOME_SCFSKP2_MEDIATED_DEGRADATION_OF_P27_P21 | 46 | 0.43 | 1.61 | 0.014 | 0.093 |
| 33 | KEGG_ANTIGEN_PROCESSING_AND_PRESENTATION | 24 | 0.50 | 1.61 | 0.015 | 0.092 |
| 34 | REACTOME_AUTODEGRADATION_OF_THE_E3_UBIQUITIN_LIGASE_COP1 | 44 | 0.43 | 1.60 | 0.011 | 0.094 |
| 35 | REACTOME_P53_INDEPENDENT_G1_S_DNA_DAMAGE_CHECKPOINT | 44 | 0.43 | 1.60 | 0.014 | 0.096 |
| 36 | REACTOME_VIF_MEDIATED_DEGRADATION_OF_APOBEC3G | 46 | 0.42 | 1.60 | 0.015 | 0.094 |
| 37 | REACTOME_SIGNALING_BY_THE_B_CELL_RECEPTOR_BCR | 72 | 0.38 | 1.59 | 0.006 | 0.100 |
| 38 | REACTOME_METABOLISM_OF_PROTEINS | 175 | 0.34 | 1.59 | 0.001 | 0.098 |
| 39 | REACTOME_CYTOKINE_SIGNALING_IN_IMMUNE_SYSTEM | 79 | 0.37 | 1.57 | 0.013 | 0.105 |
| 40 | REACTOME_CYCLIN_E_ASSOCIATED_EVENTS_DURING_G1_S_TRANSITION_ | 48 | 0.42 | 1.57 | 0.019 | 0.102 |
| 41 | REACTOME_ER_PHAGOSOME_PATHWAY | 50 | 0.41 | 1.56 | 0.016 | 0.112 |
| 42 | REACTOME_CROSS_PRESENTATION_OF_SOLUBLE_EXOGENOUS_ANTIGENS_ENDOSOMES | 42 | 0.42 | 1.55 | 0.017 | 0.119 |
| 43 | REACTOME_SYNTHESIS_OF_DNA | 57 | 0.39 | 1.55 | 0.014 | 0.117 |

|  |  |  |  |  |  |  |
|---|---|---|---|---|---|---|
| 44 | REACTOME_ASSEMBLY_OF_THE_PRE_REPLICATIVE_COMPLEX | 48 | 0.41 | 1.55 | 0.025 | 0.114 |
| 45 | REACTOME_REGULATION_OF_APOPTOSIS | 45 | 0.41 | 1.53 | 0.020 | 0.123 |
| 46 | REACTOME_CELL_CYCLE_CHECKPOINTS | 64 | 0.38 | 1.53 | 0.014 | 0.126 |
| 47 | REACTOME_SCF_BETA_TRCP_MEDIATED_DEGRADATION_OF_EMI1 | 44 | 0.41 | 1.53 | 0.022 | 0.124 |
| 48 | REACTOME_APC_C_CDC20_MEDIATED_DEGRADATION_OF_MITOTIC_PROTEINS | 50 | 0.40 | 1.52 | 0.021 | 0.122 |
| 49 | REACTOME_HOST_INTERACTIONS_OF_HIV_FACTORS | 76 | 0.36 | 1.51 | 0.012 | 0.135 |
| 50 | REACTOME_APC_C_CDH1_MEDIATED_DEGRADATION_OF_CDC20_AND_OTHER_APC_C_CDH1_TARGETED_PROTEINS_IN_LATE_MITOSIS_EARLY_G1 | 50 | 0.39 | 1.49 | 0.021 | 0.148 |
| 51 | REACTOME_FACTORS_INVOLVED_IN_MEGAKARYOCYTE_DEVELOPMENT_AND_PLATELET_PRODUCTION | 29 | 0.43 | 1.49 | 0.030 | 0.145 |
| 52 | REACTOME_PI_3K_CASCADE | 16 | 0.50 | 1.46 | 0.045 | 0.164 |
| 53 | REACTOME_SIGNALING_BY_WNT | 51 | 0.38 | 1.46 | 0.039 | 0.162 |
| 54 | REACTOME_M_G1_TRANSITION | 50 | 0.37 | 1.42 | 0.045 | 0.197 |
| 55 | REACTOME_APOPTOSIS | 72 | 0.34 | 1.41 | 0.045 | 0.215 |
| 56 | REACTOME_IMMUNE_SYSTEM | 293 | 0.27 | 1.38 | 0.014 | 0.230 |

**Supplemental Table 6. Gene sets enriched in phenotype HJ compared to J (Erythroblasts).**

| | GS | SIZE | ES | NES | NOM p-val | FDR q-val |
|---|---|---|---|---|---|---|
| 1 | REACTOME_NONSENSE_MEDIATED_DECAY_ENHANCED_BY_THE_EXON_JUNCTION_COMPLEX | 88 | 0.53 | 2.20 | 0.000 | 0.002 |
| 2 | REACTOME_3_UTR_MEDIATED_TRANSLATIONAL_REGULATION | 82 | 0.53 | 2.18 | 0.000 | 0.002 |
| 3 | REACTOME_PEPTIDE_CHAIN_ELONGATION | 76 | 0.54 | 2.17 | 0.000 | 0.002 |
| 4 | REACTOME_ACTIVATION_OF_THE_MRNA_UPON_BINDING_OF_THE_CAP_BINDING_COMPLEX_AND_EIFS_AND_SUBSEQUENT_BINDING_TO_43S | 40 | 0.59 | 2.12 | 0.000 | 0.002 |
| 5 | REACTOME_FORMATION_OF_THE_TERNARY_COMPLEX_AND_SUBSEQUENTLY_THE_43S_COMPLEX | 34 | 0.62 | 2.10 | 0.000 | 0.002 |
| 6 | REACTOME_METABOLISM_OF_MRNA | 149 | 0.46 | 2.07 | 0.000 | 0.004 |
| 7 | KEGG_RIBOSOME | 76 | 0.50 | 2.02 | 0.000 | 0.007 |
| 8 | REACTOME_METABOLISM_OF_RNA | 162 | 0.44 | 2.01 | 0.000 | 0.006 |
| 9 | REACTOME_TRANSLATION | 103 | 0.46 | 1.96 | 0.000 | 0.012 |
| 10 | REACTOME_SRP_DEPENDENT_COTRANSLATIONAL_PROTEIN_TARGETING_TO_MEMBRANE | 87 | 0.46 | 1.88 | 0.000 | 0.026 |
| 11 | REACTOME_INFLUENZA_VIRAL_RNA_TRANSCRIPTION_AND_REPLICATION | 81 | 0.46 | 1.87 | 0.000 | 0.025 |
| 12 | REACTOME_INFLUENZA_LIFE_CYCLE | 93 | 0.44 | 1.82 | 0.000 | 0.040 |
| 13 | REACTOME_P53_DEPENDENT_G1_DNA_DAMAGE_RESPONSE | 47 | 0.49 | 1.80 | 0.000 | 0.044 |
| 14 | REACTOME_ORC1_REMOVAL_FROM_CHROMATIN | 51 | 0.46 | 1.72 | 0.002 | 0.097 |
| 15 | REACTOME_AUTODEGRADATION_OF_THE_E3_UBIQUITIN_LIGASE_COP1 | 44 | 0.47 | 1.72 | 0.005 | 0.093 |
| 16 | REACTOME_CDT1_ASSOCIATION_WITH_THE_CDC6_ORC_ORIGIN_COMPLEX | 43 | 0.46 | 1.72 | 0.007 | 0.090 |
| 17 | REACTOME_M_G1_TRANSITION | 50 | 0.46 | 1.70 | 0.000 | 0.096 |
| 18 | REACTOME_SYNTHESIS_OF_DNA | 57 | 0.44 | 1.70 | 0.003 | 0.092 |
| 19 | REACTOME_ASSEMBLY_OF_THE_PRE_REPLICATIVE_COMPLEX | 48 | 0.46 | 1.69 | 0.003 | 0.094 |
| 20 | REACTOME_REGULATION_OF_APOPTOSIS | 45 | 0.46 | 1.69 | 0.007 | 0.096 |
| 21 | KEGG_PROTEASOME | 41 | 0.47 | 1.68 | 0.000 | 0.093 |

|  |  |  |  |  |  |  |
|---|---|---|---|---|---|---|
| 22 | REACTOME_CDK_MEDIATED_PHOSPHORYLATION_AND_REMOVAL_OF_CDC6 | 43 | 0.46 | 1.68 | 0.007 | 0.090 |
| 23 | REACTOME_ACTIVATION_OF_NF_KAPPAB_IN_B_CELLS | 50 | 0.45 | 1.67 | 0.007 | 0.097 |
| 24 | REACTOME_METABOLISM_OF_PROTEINS | 175 | 0.36 | 1.66 | 0.000 | 0.100 |
| 25 | REACTOME_DOWNSTREAM_SIGNALING_EVENTS_OF_B_CELL_RECEPTOR_BCR | 64 | 0.42 | 1.65 | 0.003 | 0.106 |
| 26 | REACTOME_SCFSKP2_MEDIATED_DEGRADATION_OF_P27_P21 | 46 | 0.44 | 1.65 | 0.010 | 0.105 |
| 27 | REACTOME_DNA_REPLICATION | 85 | 0.40 | 1.64 | 0.007 | 0.107 |
| 28 | REACTOME_MITOTIC_M_M_G1_PHASES | 76 | 0.41 | 1.64 | 0.005 | 0.105 |
| 29 | REACTOME_P53_INDEPENDENT_G1_S_DNA_DAMAGE_CHECKPOINT | 44 | 0.44 | 1.63 | 0.005 | 0.106 |
| 30 | REACTOME_SIGNALING_BY_THE_B_CELL_RECEPTOR_BCR | 72 | 0.40 | 1.62 | 0.005 | 0.109 |
| 31 | REACTOME_VIF_MEDIATED_DEGRADATION_OF_APOBEC3G | 46 | 0.44 | 1.62 | 0.010 | 0.114 |
| 32 | REACTOME_DESTABILIZATION_OF_MRNA_BY_AUF1_HNRNP_D0 | 46 | 0.44 | 1.60 | 0.010 | 0.120 |
| 33 | REACTOME_REGULATION_OF_MRNA_STABILITY_BY_PROTEINS_THAT_BIND_AU_RICH_ELEMENTS | 56 | 0.42 | 1.60 | 0.005 | 0.120 |
| 34 | REACTOME_AUTODEGRADATION_OF_CDH1_BY_CDH1_APC_C | 47 | 0.43 | 1.60 | 0.007 | 0.118 |
| 35 | PID_SMAD2_3NUCLEAR_PATHWAY | 28 | 0.47 | 1.57 | 0.023 | 0.145 |
| 36 | REACTOME_G1_S_TRANSITION | 60 | 0.41 | 1.57 | 0.013 | 0.142 |
| 37 | REACTOME_REGULATION_OF_ORNITHINE_DECARBOXYLASE_ODC | 46 | 0.42 | 1.56 | 0.011 | 0.142 |
| 38 | PID_E2F_PATHWAY | 26 | 0.47 | 1.56 | 0.021 | 0.138 |
| 39 | REACTOME_SCF_BETA_TRCP_MEDIATED_DEGRADATION_OF_EMI1 | 44 | 0.44 | 1.56 | 0.015 | 0.135 |
| 40 | REACTOME_ER_PHAGOSOME_PATHWAY | 50 | 0.42 | 1.56 | 0.016 | 0.136 |
| 41 | REACTOME_CYCLIN_E_ASSOCIATED_EVENTS_DURING_G1_S_TRANSITION_ | 48 | 0.42 | 1.56 | 0.012 | 0.134 |
| 42 | REACTOME_S_PHASE | 61 | 0.40 | 1.56 | 0.015 | 0.132 |
| 43 | PID_IL4_2PATHWAY | 16 | 0.55 | 1.55 | 0.040 | 0.133 |
| 44 | REACTOME_SIGNALING_BY_WNT | 51 | 0.41 | 1.55 | 0.009 | 0.137 |
| 45 | BIOCARTA_PROTEASOME_PATHWAY | 26 | 0.47 | 1.54 | 0.026 | 0.140 |
| 46 | PID_P73PATHWAY | 21 | 0.49 | 1.54 | 0.039 | 0.140 |

|  |  |  |  |  |  |  |
|---|---|---|---|---|---|---|
| 47 | REACTOME_CELL_CYCLE_CHECKPOINTS | 64 | 0.39 | 1.53 | 0.015 | 0.142 |
| 48 | REACTOME_CROSS_PRESENTATION_OF_SOLUBLE_EXOGENOUS_ANTIGENS_ENDOSOMES | 42 | 0.42 | 1.52 | 0.021 | 0.155 |
| 49 | REACTOME_MITOTIC_G1_G1_S_PHASES | 70 | 0.38 | 1.51 | 0.019 | 0.157 |
| 50 | BIOCARTA_CHREBP2_PATHWAY | 16 | 0.53 | 1.50 | 0.042 | 0.171 |
| 51 | PID_FCER1_PATHWAY | 21 | 0.48 | 1.46 | 0.041 | 0.219 |
| 52 | REACTOME_METABOLISM_OF_LIPIDS_AND_LIPOPROTEINS | 94 | 0.35 | 1.44 | 0.019 | 0.246 |
| 53 | REACTOME_APC_C_CDH1_MEDIATED_DEGRADATION_OF_CDC20_AND_OTHER_APC_C_CDH1_TARGETED_PROTEINS_IN_LATE_MITOSIS_EARLY_G1 | 50 | 0.39 | 1.43 | 0.048 | 0.247 |

**Supplemental Table 7. Mutations detected in the patients.**

| Case | Diagnosis | *MIRlet-7c* | *HMGA2* mRNA | Gene | Codon change | Amino acid change |
|---|---|---|---|---|---|---|
| S017 | ET | >2.00 | <0.10 | DNMT3A | c.2645G>A | p.R882H |
| | | | | JAK2 | c.1849G>T | p.V617F |
| S018 | ET | 0.41 | <0.10 | JAK2 | c.1849G>T | p.V617F |
| | | | | TET2 | c.5696T>G | p.L1899R |
| S019 | ET | >2.00 | <0.10 | CALR | insertion 5 | |
| S024 | ET | 1.33 | 0.55 | JAK2 | c.1849G>T | p.V617F |
| S034 | ET | 1.38 | 0.30 | JAK2 | c.1849G>T | p.V617F |
| S036 | ET | **0.13** | 0.78 | JAK2 | c.1849G>T | p.V617F |
| | | | | PRPF40B | c.2533C>T | p.Q845* |
| | | | | PRPF40B | c.2537A>T | p.Q846L |
| | | | | PRPF40B | c.2539G>T | p.A847S |
| S043 | ET | **0.17** | **1.78** | JAK2 | c.1849G>T | p.V617F |
| S044 | ET | **0.17** | 0.54 | JAK2 | c.1849G>T | p.V617F |
| S050 | ET | **0.28** | **1.50** | JAK2 | c.1849G>T | p.V617F |
| S051 | ET | 0.78 | 0.78 | DNMT3A | c.2645G>A | p.R882H |
| | | | | JAK2 | c.1849G>T | p.V617F |
| S075 | ET | **0.29** | **1.89** | none | | |
| S077 | ET | **0.01** | 0.47 | CALR | deletion 4 | |
| | | | | SF3B1 | c.1996A>C | p.K666Q |
| S078 | ET | **0.07** | **39.74** | JAK2 | c.1849G>T | p.V617F |
| S090 | ET | **0.02** | 0.46 | JAK2 | c.1849G>T | p.V617F |
| | | | | PRPF40B | | p.D?L |
| | | | | TET2 | | p.-274 |
| | | | | TP53 | c.815T>A | p.V272E |
| S091 | ET | **0.02** | **2.78** | JAK2 | c.1849G>T | p.V617F |
| | | | | TET2 | | p.SF1284S |
| S095 | ET | 0.37 | 0.42 | JAK2 | c.1849G>T | p.V617F |
| S107 | ET | **0.16** | **1.03** | JAK2 | c.1849G>T | p.V617F |
| S117 | ET | **0.03** | **2.74** | JAK2 | c.1849G>T | p.V617F |
| | | | | TET2 | c.1249C>T | p.Q417* |
| S118 | ET | 1.39 | 0.27 | JAK2 | c.1849G>T | p.V617F |
| S119 | ET | 0.49 | 0.68 | JAK2 | c.1849G>T | p.V617F |
| S1002 | ET | 0.33 | **2.55** | JAK2 | c.1849G>T | p.V617F |
| S1004 | ET | 0.52 | 0.62 | JAK2 | c.1849G>T | p.V617F |

| | | | | | | |
|---|---|---|---|---|---|---|
| | | | | TET2 | | p.-848Y? |
| S1005 | ET | 1.33 | <0.10 | JAK2 | c.1849G>T | p.V617F |
| S1008 | ET | 0.66 | 0.63 | JAK2 | c.1849G>T | p.V617F |
| S1009 | ET | 1.29 | 0.92 | none | | |
| S1019 | ET | **0.06** | **1.92** | JAK2 | c.1849G>T | p.V617F |
| | | | | JAK2 | c.2569A>G | p.K857E |
| S1105 | ET | **0.23** | <0.10 | JAK2 | c.1849G>T | p.V617F |
| S023 | PMF | 0.36 | **101.99** | CBL | c.1199T>G | p.M400R |
| | | | | IDH1 | c.395G>A | p.R132H |
| | | | | JAK2 | c.1849G>T | p.V617F |
| S037 | PMF | 0.73 | **82.56** | ASXL1 | c.2077C>T | p.R693* |
| | | | | JAK2 | c.1849G>T | p.V617F |
| S045 | PMF | 0.36 | **1.56** | JAK2 | c.1849G>T | p.V617F |
| S046 | PMF | 0.47 | **1.70** | ASXL1 | | p.-591? |
| | | | | EZH2 | | p.-165 |
| | | | | JAK2 | c.1849G>T | p.V617F |
| S053 | PMF | **0.26** | **3.52** | IDH2 | c.419G>A | p.R140Q |
| | | | | KRAS | c.182A>G | p.Q61R |
| | | | | RUNX1 | c.205G>C | p.G69R |
| S093 | PMF | 1.01 | **2.53** | none | | |
| S094 | PMF | **0.06** | 0.55 | JAK2 | c.1849G>T | p.V617F |
| S114 | PMF | 0.36 | **13.79** | JAK2 | c.1849G>T | p.V617F |
| | | | | SF3B1 | c.1998G>C | p.K666N |
| S120 | PMF | 0.41 | **1.06** | JAK2 | c.1849G>T | p.V617F |
| | | | | EZH2 | c.1981A>G | p.K661E |
| S127 | PMF | **0.22** | **7.83** | CALR | deletion 52 | |
| | | | | KRAS | c.35G>C | p.G12A |
| | | | | SF3B1 | c.1998G>T | p.K666N |
| S1015 | PMF | 0.77 | **25.21** | SETBP1 | c.2602G>A | p.D868N |
| | | | | STAG2 | c.1042C>T | p.L348F |
| S1016 | PMF | **0.18** | **9.43** | JAK2 | c.1849G>T | p.V617F |
| | | | | SF3B1 | c.1996A>C | p.K666Q |
| S1023 | PMF | **0.09** | **7.19** | JAK2 | c.1849G>T | p.V617F |
| | | | | KRAS | c.35G>C | p.G12A |
| S1106 | PMF | 0.34 | **1.63** | DNMT3A | c.2644C>T | p.R882C |
| | | | | MPL | | p.W515L |
| | | | | TET2 | c.4075C>T | p.R1359C |

| | | | | | | |
|---|---|---|---|---|---|---|
| S1137 | PMF | 1.26 | **255.50** | ASXL1 | c.2548G>T | p.E850* |
| | | | | JAK2 | c.1849G>T | p.V617F |
| | | | | SMC3 | | p.LK933L |
| S1145 | PMF | 0.83 | **23.93** | JAK2 | c.1849G>T | p.V617F |
| | | | | SF3A1 | | p.N?T |
| | | | | TET2 | c.3851C>T | p.S1284F |
| S1146 | PMF | **0.09** | **39.28** | JAK2 | c.1849G>T | p.V617F |
| | | | | TET2 | c.3319T>C | p.S1107P |
| | | | | TP53 | c.91G>A | p.V31I |
| S1151 | PMF | **0.03** | **1.85** | ASXL1 | c.2548G>T | p.E850* |
| | | | | IDH1 | c.38T>A | p.M13K |
| | | | | JAK2 | c.1849G>T | p.V617F |
| | | | | TET2 | c.3319T>C | p.S1107P |

Low *MIRlet-7* and high *HMGA2* mRNA levels are indicated as blue and red, respectively.