|  |
| --- |
| **Table e-6:** Reactome gene ontology pathways for behavioral variant frontotemporal dementia |
| **Reactome pathways** | **Reference list** | **# Genes overlapping** | **# Genes expected** | **Fold Enrichment** | **P-value** |
| Signaling by EGFRvIII in Cancer (R-HSA-5637812) | 14 | 4 | 0.04 | 96.65 | 1.91E-04 |
| Constitutive Signaling by EGFRvIII (R-HSA-5637810) | 14 | 4 | 0.04 | 96.65 | 1.91E-04 |
| Constitutive Signaling by Ligand-Responsive EGFR Cancer Variants (R-HSA-1236382) | 18 | 4 | 0.05 | 75.17 | 5.16E-04 |
| Signaling by EGFR in Cancer (R-HSA-1643713) | 18 | 4 | 0.05 | 75.17 | 5.16E-04 |
| Signaling by Ligand-Responsive EGFR Variants in Cancer (R-HSA-5637815) | 18 | 4 | 0.05 | 75.17 | 5.16E-04 |
| Signaling by ERBB2 (R-HSA-1227986) | 43 | 4 | 0.13 | 31.47 | 1.59E-02 |
| GPVI-mediated activation cascade (R-HSA-114604) | 55 | 4 | 0.16 | 24.6 | 4.15E-02 |
| Cyclin A:Cdk2-associated events at S phase entry (R-HSA-69656) | 70 | 5 | 0.21 | 24.16 | 4.06E-03 |
| Regulation of actin dynamics for phagocytic cup formation (R-HSA-2029482) | 90 | 5 | 0.27 | 18.79 | 1.36E-02 |
| Fcgamma receptor (FCGR) dependent phagocytosis (R-HSA-2029480) | 114 | 6 | 0.34 | 17.8 | 2.17E-03 |
| S Phase (R-HSA-69242) | 128 | 6 | 0.38 | 15.86 | 4.21E-03 |
| G1/S Transition (R-HSA-69206) | 115 | 5 | 0.34 | 14.71 | 4.39E-02 |
| Mitotic G1-G1/S phases (R-HSA-453279) | 139 | 6 | 0.41 | 14.6 | 6.73E-03 |
| Interleukin-3, 5 and GM-CSF signaling (R-HSA-512988) | 250 | 8 | 0.74 | 10.82 | 1.38E-03 |
| Diseases of signal transduction (R-HSA-5663202) | 282 | 9 | 0.83 | 10.8 | 2.72E-04 |
| MAPK1/MAPK3 signaling (R-HSA-5684996) | 229 | 7 | 0.68 | 10.34 | 9.55E-03 |
| MAPK family signaling cascades (R-HSA-5683057) | 267 | 8 | 0.79 | 10.14 | 2.25E-03 |
| VEGFA-VEGFR2 Pathway (R-HSA-4420097) | 307 | 9 | 0.91 | 9.92 | 5.52E-04 |
| FCERI mediated MAPK activation (R-HSA-2871796) | 276 | 8 | 0.82 | 9.8 | 2.87E-03 |
| Signaling by SCF-KIT (R-HSA-1433557) | 311 | 9 | 0.92 | 9.79 | 6.14E-04 |
| Signaling by VEGF (R-HSA-194138) | 316 | 9 | 0.93 | 9.63 | 7.01E-04 |
| Signaling by Interleukins (R-HSA-449147) | 386 | 10 | 1.14 | 8.76 | 3.55E-04 |
| Signaling by ERBB4 (R-HSA-1236394) | 315 | 8 | 0.93 | 8.59 | 7.55E-03 |
| Signaling by FGFR4 (R-HSA-5654743) | 318 | 8 | 0.94 | 8.51 | 8.09E-03 |
| Signaling by FGFR3 (R-HSA-5654741) | 319 | 8 | 0.94 | 8.48 | 8.27E-03 |
| Signaling by FGFR1 (R-HSA-5654736) | 323 | 8 | 0.95 | 8.38 | 9.06E-03 |
| DAP12 signaling (R-HSA-2424491) | 330 | 8 | 0.98 | 8.2 | 1.06E-02 |
| Fc epsilon receptor (FCERI) signaling (R-HSA-2454202) | 376 | 9 | 1.11 | 8.1 | 2.92E-03 |
| Signaling by EGFR (R-HSA-177929) | 336 | 8 | 0.99 | 8.05 | 1.21E-02 |
| DAP12 interactions (R-HSA-2172127) | 344 | 8 | 1.02 | 7.87 | 1.43E-02 |
| Signaling by FGFR2 (R-HSA-5654738) | 347 | 8 | 1.03 | 7.8 | 1.52E-02 |
| Signaling by FGFR (R-HSA-190236) | 353 | 8 | 1.04 | 7.67 | 1.72E-02 |
| Cell Cycle, Mitotic (R-HSA-69278) | 471 | 9 | 1.39 | 6.46 | 1.78E-02 |
| Cytokine Signaling in Immune system (R-HSA-1280215) | 610 | 11 | 1.8 | 6.1 | 2.89E-03 |
| Innate Immune System (R-HSA-168249) | 790 | 13 | 2.34 | 5.57 | 7.99E-04 |
| Disease (R-HSA-1643685) | 899 | 13 | 2.66 | 4.89 | 3.36E-03 |
| Developmental Biology (R-HSA-1266738) | 806 | 11 | 2.38 | 4.62 | 3.96E-02 |
| Immune System (R-HSA-168256) | 1604 | 17 | 4.74 | 3.59 | 4.81E-03 |
| Signal Transduction (R-HSA-162582) | 2460 | 25 | 7.27 | 3.44 | 1.72E-05 |
| Unclassified (UNCLASSIFIED) | 11676 | 11 | 34.52 | 0.32 | 0.00E+00 |

**Table e-6 Legend.**  Reactome pathways highlighted in our ontological analysis and corresponding pathway reference numbers. Reference list – number of genes documented in the Reactome database for each pathway, # Genes overlapping – number of genes in both pathway reference list and set of 62 enriched bvFTD genes, # Genes expected – Reactome a priori expected degree of overlap given number of genes entered into the pathway analysis tool, Fold enrichment – Reactome degree of enrichment based on expected value and number of overlapping genes, p value – Bonferroni corrected p values.