

Supplementary Table 6. Significantly enriched disease classes using 92 gene list differentially expressed between germline MMR deficient and assumed somatic MMR deficient endometrioid tumors

Category	Term	Fold enrichment	p-value	Benjamini	Genes
<i>GAD_DISEASE_CLASS</i>	<i>METABOLIC</i>	<i>1.51</i>	<i>7.82E-04</i>	0.01	<i>CSF3, CYP24A1, CYP3A5, PDLIM5, SSFA2, OBP2B, CLU, POMC, MEGF11, PGBD5, COL9A1, SAA2, SAA1, BCL11A, LRRTM1, COL6A1, LRRC55, ZC3H12C, PITX2, GABRG3, ATRNL1, CHI3L2, C4BPA, CACNA1S, THSD7B, DKK4, C1QTNF9B, PGPEP1L, CEACAM22P, GRM6, TFAP2B, FAM155A, GPA33, CYP2A6, C2CD4A, ZNF385B, HAS3, UTP20, ATP6V0A4, PDZK1, KIF26B</i>
GAD_DISEASE_CLASS	DEVELOPMENTAL	2.06	7.42E-03	0.06	<i>CYP3A5, GABRG3, ESRRB, POMC, ZIC1, CACNA1S, COL9A1, NEDD4, LRRTM1, FAM155A, CYP2A6, TUBA4A, ZNF385B, C2CD4A, PITX2, KIF26B</i>
GAD_DISEASE_CLASS	CHEMDEPENDENCY	1.54	8.41E-03	0.05	<i>CPLX3, POMC, KANK1, SEMA5B, COL9A1, PECR, SAA2, BCL11A, LRRTM1, COL6A1, LRRC55, PAPSS1, ZC3H12C, PITX2, GABRG3, ATRNL1, ESRRB, CHI3L2, CACNA1S, THSD7B, GRM6, HIVEP3, TFAP2B, GPA33, FAM155A, CYP2A6, ZNF385B, ATP6V0A4, KIF26B</i>
GAD_DISEASE_CLASS	PSYCH	1.77	0.02	0.08	<i>CPLX3, CYP3A5, GABRG3, CES1, ATRNL1, PDLIM5, CLU, POMC, DKK4, PGBD5, SAA2, BCL11A, GRM6, TFAP2B, FAM155A, CYP2A6, ZNF385B, HAS3</i>
GAD_DISEASE_CLASS	CARDIOVASCULAR	1.41	0.02	0.08	<i>CSF3, CYP24A1, CYP3A5, CPLX3, CLU, CX3CL1, DEFB4A, POMC, MEGF11, PGBD5, COL9A1, PECR, SAA2, SAA1, LRRC55, PAPSS1, ZC3H12C, PITX2, C9ORF66, GABRG3, ATRNL1, THSD7B, TNNT2, NEDD4, HIVEP3, GPA33, FAM155A, ZNF385B, UTP20, PDZK1, KIF26B</i>
GAD_DISEASE_CLASS	UNKNOWN	1.80	0.03	0.09	<i>CYP3A5, CYP24A1, GABRG3, CES1, MLH1, POMC, CACNA1S, TNNT2, SAA1, NEDD4, BCL11A, PI3, TFAP2B, CYP2A6, COL6A1</i>
GAD_DISEASE_CLASS	NEUROLOGICAL	1.47	0.05	0.12	<i>CYP3A5, GABRG3, ATRNL1, CLU, CHI3L2, CX3CL1, POMC, CACNA1S, THSD7B, SAA2, SAA1, LRRTM1, HIVEP3, TFAP2B, FAM155A, CYP2A6, COL6A1, ZNF385B, C2CD4A, PAPSS1, PITX2, KIF26B</i>
GAD_DISEASE_CLASS	PHARMACOGENOMIC	1.50	0.05	0.10	<i>CSF3, CYP24A1, CYP3A5, CES1, CX3CL1, POMC, DKK4, PDZK1IP1, GPX2, TNNT2, COL9A1, SEMA5B, SAA2, SAA1, BCL11A, HIVEP3, TFAP2B, CYP2A6, HAS3, PDZK1, PITX2</i>
GAD_DISEASE_CLASS	OTHER	1.70	0.06	0.11	<i>CSF3, CYP3A5, CYP24A1, MLH1, DEFB4A, POMC, PRKY, GPX2, COL9A1, LRRTM1, FAM155A, CYP2A6, COL6A1, PITX2</i>
GAD_DISEASE_CLASS	CANCER	1.33	0.11	0.20	<i>CSF3, CYP24A1, CYP3A5, CES1, TMEM213, ESRRB, PDLIM5, CLU, MLH1, C4BPA, CX3CL1, POMC, MEGF11, DKK4, EPM2AIP1, GPX2, SEMA5B, NEDD4, DEFA5, LRRTM1, CYP2A6, PAPSS1</i>
GAD_DISEASE_CLASS	HEMATOLOGICAL	1.39	0.25	0.37	<i>CSF3, CYP3A5, ATRNL1, SAA1, ESRRB, BCL11A, LRRTM1, FAM155A, C4BPA, ATP6V0A4, LGR6</i>
GAD_DISEASE_CLASS	AGING	1.43	0.35	0.48	<i>CYP3A5, SAA2, ATRNL1, PDLIM5, HIVEP3, MLH1, THSD7B</i>
GAD_DISEASE_CLASS	REPRODUCTION	1.41	0.36	0.46	<i>CSF3, CYP3A5, DEFA5, LRRTM1, TFAP2B, POMC, THSD7B</i>
GAD_DISEASE_CLASS	VISION	1.40	0.47	0.56	<i>GRM6, CLU, C4BPA, ATP6V0A4, PITX2</i>

Significantly enriched classes are in bold italics (adjusted p-value <0.05 used as significance cut-off)