

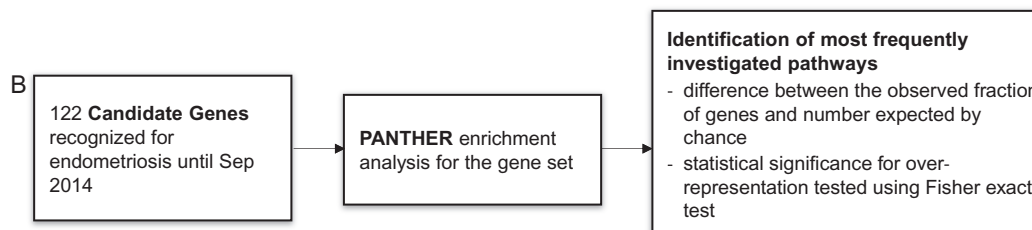
A

GWAS
 International EndoGene Consortium (IEC)
 3,194 cases and 7,060 controls
 Analysis tier
 Overall vs. controls
 Stage A vs. controls
 Stage B vs. controls

MSigDB
 (1) Curated Canonical pathways:
 BioCarta; KEGG^a; Matrisome; PID^b;
 Reactome; SA^c; SIG^d; ST^e;
 SuperArray
 1,330 gene sets
 (2) Gene Ontology (GO) gene sets:
 1,454 gene sets
PANTHER
 149 pathways
 TOTAL 2,860 gene sets
 (gene set size >10 tested)

MAGENTA Software
 Algorithm
 STEP 1 – map SNP's onto genes
 STEP 2 – score genes based on
 regional SNP p-values
 STEP 3 – correct for gene score
 confounders
 STEP 4 – estimate gene set
 enrichment P-value for gene
 sets of interest^f

RESULTS of enriched pathways for
 (1) Overall endometriosis
 (2) Stage A endometriosis
 (3) Stage B endometriosis



Supplementary Figure S2 Study designs for (A) the GWAS pathway analysis on endometriosis and (B) identification of pathways that have been investigated indirectly through candidate gene association studies for endometriosis. ^aKyoto Encyclopedia of Genes and Genomes; ^bPathway Interaction Database; ^cSigmaAldrich; ^dSignaling Gateway; ^eSignal Transduction; ^fSegre *et al. PLoS Genetics*, 2010.