

Supplementary Table 4. Comparison of enriched gene sets associated with lung function as identified by two programs: iGSEA4GWAS vs. GSA-SNP. For each algorithm, enriched pathways were required to be significantly associated with at least one lung function measure in both CHARGE and SpiroMeta at FDR < 0.05. Since GSA-SNP sampled the full GO database, we show only representative categories for similar processes.

Glycoprotein

iGSEA4GWAS	GSA-SNP
HSA00534 HEPARAN SULFATE BIOSYNTHESIS	HSA00534 HEPARAN SULFATE BIOSYNTHESIS
CHONDROITIN	HEPARAN SULFATE PROTEOGLYCAN BINDING
HEPARAN SULFATE BIOSYNTHESIS	HEPARAN SULFATE PROTEOGLYCAN BIOSYNTHETIC PROCESS
PROTEOGLYCAN BIOSYNTHETIC PROCESS	HEPARAN SULFATE PROTEOGLYCAN METABOLIC PROCESS
	HEPARIN BINDING
	GLYCOSAMINOGLYCAN BINDING
	PROTEOGLYCAN BINDING
	PROTEOGLYCAN METABOLIC PROCESS

Metabolism

iGSEA4GWAS	GSA-SNP
HSA00062 FATTY ACID ELONGATION IN MITOCHONDRIA	HSA00062 FATTY ACID ELONGATION IN MITOCHONDRIA
CARBOHYDRATE KINASE ACTIVITY	CARBOHYDRATE BINDING
SULFOTRANSFERASE ACTIVITY	ACETYLGALACTOSAMINYLTRANSFERASE ACTIVITY
OXIDOREDUCTASE ACTIVITY ACTING ON SULFUR GROUP OF DONORS	POLYPEPTIDE N ACETYLGALACTOSAMINYLTRANSFERASE ACTIVITY
ETCPATHWAY	POSITIVE REGULATION OF NUCLEOTIDE METABOLIC PROCESS
CARBOXY LYASE ACTIVITY	POSITIVE REGULATION OF PURINE NUCLEOTIDE METABOLIC PROCESS
HISTIDINE METABOLISM	REGULATION OF HORMONE METABOLIC PROCESS
	REGULATION OF NUCLEOTIDE METABOLIC PROCESS
	CAMP METABOLIC PROCESS

Channel/Transporter

iGSEA4GWAS

CHANNEL REGULATOR ACTIVITY
POTASSIUM ION TRANSPORT
POTASSIUM CHANNEL ACTIVITY
VOLTAGE GATED CALCIUM CHANNEL ACTIVITY
ANTIPORTER ACTIVITY
REGULATION OF HEART CONTRACTION
AUXILIARY TRANSPORT PROTEIN ACTIVITY
CATION TRANSMEMBRANE TRANSPORTER ACTIVITY
SUBSTRATE SPECIFIC TRANSPORTER ACTIVITY
CATION TRANSPORTING ATPASE ACTIVITY
PHOSPHOLIPID TRANSPORTER ACTIVITY
LIPID TRANSPORTER ACTIVITY

GSA-SNP

CHANNEL REGULATOR ACTIVITY
POTASSIUM ION TRANSPORT
POTASSIUM CHANNEL ACTIVITY
VOLTAGE GATED CALCIUM CHANNEL ACTIVITY
VOLTAGE GATED CATION CHANNEL ACTIVITY
VOLTAGE GATED CHANNEL ACTIVITY
VOLTAGE GATED ION CHANNEL ACTIVITY
VOLTAGE GATED POTASSIUM CHANNEL ACTIVITY
CALCIUM ION TRANSMEMBRANE TRANSPORT
CALCIUM ION TRANSMEMBRANE TRANSPORTER ACTIVITY
CALCIUM ION TRANSPORT
CALCIUM CHANNEL ACTIVITY
CATION CHANNEL ACTIVITY
CALCIUM RELEASE CHANNEL ACTIVITY
CATION CHANNEL COMPLEX
CATION HOMEOSTASIS
CATION TRANSPORT
CHANNEL ACTIVITY
CHANNEL REGULATOR ACTIVITY
EXCITATORY EXTRACELLULAR LIGAND GATED ION CHANNEL ACTIVITY
GATED CHANNEL ACTIVITY
ION CHANNEL ACTIVITY
ION CHANNEL COMPLEX
ACTIVE TRANSMEMBRANE TRANSPORTER ACTIVITY
ATPASE ACTIVITY
ATPASE ACTIVITY COUPLED
ATPASE ACTIVITY COUPLED TO TRANSMEMBRANE MOVEMENT OF SUBSTANCES
PHOSPHOLIPID TRANSLOCATING ATPASE ACTIVITY
LIPID TRANSPORT

Cell Singaling

igSEA4GWAS	GSA-SNP
HSA04020 CALCIUM SIGNALING PATHWAY	HSA04020 CALCIUM SIGNALING PATHWAY
G PROTEIN SIGNALING COUPLED TO CAMP NUCLEOTIDE SECOND MESSENGER	G PROTEIN SIGNALING COUPLED TO CAMP NUCLEOTIDE SECOND MESSENGER
G PROTEIN SIGNALING COUPLED TO CYCLIC NUCLEOTIDE SECOND MESSENGER	G PROTEIN SIGNALING COUPLED TO CYCLIC NUCLEOTIDE SECOND MESSENGER
HSA04080 NEUROACTIVE LIGAND RECEPTOR INTERACTION	HSA04080 NEUROACTIVE LIGAND RECEPTOR INTERACTION
SECOND MESSENGER MEDIATED SIGNALING	SECOND MESSENGER MEDIATED SIGNALING
TRANSMEMBRANE RECEPTOR PROTEIN KINASE ACTIVITY	TRANSMEMBRANE RECEPTOR PROTEIN KINASE ACTIVITY
TRANSMEMBRANE RECEPTOR PROTEIN PHOSPHATASE ACTIVITY	TRANSMEMBRANE RECEPTOR PROTEIN PHOSPHATASE ACTIVITY
TRANSMEMBRANE RECEPTOR PROTEIN TYROSINE KINASE ACTIVITY	TRANSMEMBRANE RECEPTOR PROTEIN TYROSINE KINASE ACTIVITY
GPCRDB CLASS A RHODOPSIN LIKE	TRANSMEMBRANE RECEPTOR PROTEIN TYROSINE KINASE SIGNALING PATHWAY
PHOSPHOINOSITIDE MEDIATED SIGNALING	TRANSMEMBRANE RECEPTOR PROTEIN TYROSINE PHOSPHATASE ACTIVITY
PROTEIN TYROSINE KINASE ACTIVITY	PROTEIN TYROSINE KINASE ACTIVITY
ST WNT CA2 CYCLIC GMP PATHWAY	PROTEIN SERINE/THREONINE KINASE ACTIVITY
CACAMPATHWAY	ADENYLATE CYCLASE ACTIVATING G PROTEIN COUPLED RECEPTOR SIGNALING PATHWAY
GCRPATHWAY	PHOSPHATIDYLINOSITOL BINDING
PKCPATHWAY	REGULATION OF WNT RECEPTOR SIGNALING PATHWAY
TCRPATHWAY	POSITIVE REGULATION OF WNT RECEPTOR SIGNALING PATHWAY
TRKAPATHWAY	POSITIVE REGULATION OF MAPK CASCADE
PLCPATHWAY	REGULATION OF MAPK CASCADE
ACTIVATION OF NF KAPPAB TRANSCRIPTION FACTOR	T CELL RECEPTOR SIGNALING PATHWAY
POSITIVE REGULATION OF TRANSCRIPTION FACTOR ACTIVITY	POSITIVE REGULATION OF ERK1 AND ERK2 CASCADE
POSITIVE REGULATION OF DNA BINDING	POSITIVE REGULATION OF PHOSPHOLIPASE ACTIVITY
FREEPATHWAY	POSITIVE REGULATION OF PHOSPHOLIPASE C ACTIVITY
RELAPATHWAY	SEMAPHORIN PLEXIN SIGNALING PATHWAY
NTHIPATHWAY	ADENYLATE CYCLASE ACTIVATING G PROTEIN COUPLED RECEPTOR SIGNALING PATHWAY
	ADENYLATE CYCLASE MODULATING G PROTEIN COUPLED RECEPTOR SIGNALING PATHWAY
	REGULATION OF RAS PROTEIN SIGNAL TRANSDUCTION
	REGULATION OF RHO PROTEIN SIGNAL TRANSDUCTION
	REGULATION OF SMALL GTPASE MEDIATED SIGNAL TRANSDUCTION
	RAS GTPASE BINDING

RECEPTOR SIGNALING PROTEIN ACTIVITY
GTPASE ACTIVATOR ACTIVITY
GTPASE BINDING
GTPASE REGULATOR ACTIVITY

Transcription

iGSEA4GWAS

NUCLEAR REPLICATION FORK
CIRCADIANPATHWAY
HSA04710 CIRCADIAN RHYTHM
LIGAND DEPENDENT NUCLEAR RECEPTOR ACTIVITY
STEROID HORMONE RECEPTOR ACTIVITY
HISTONE MODIFICATION
ESTABLISHMENT AND OR MAINTENANCE OF CHROMATIN ARCHITECTURE
CHROMATIN MODIFICATION
THYROID HORMONE RECEPTOR BINDING
RNA POLYMERASE II TRANSCRIPTION FACTOR ACTIVITY
REGULATION OF TRANSCRIPTION DNA DEPENDENT
RIBONUCLEOPROTEIN COMPLEX
SMALL NUCLEAR RIBONUCLEOPROTEIN COMPLEX
RNA SPLICING FACTOR ACTIVITY TRANSESTERIFICATION MECHANISM
NUCLEAR SPECK
CELLULAR RESPONSE TO STRESS

GSA-SNP

TRANSCRIPTION FROM RNA POLYMERASE II PROMOTER
TRANSCRIPTION INITIATION FROM RNA POLYMERASE II PROMOTER
TRANSCRIPTION INITIATION DNA DEPENDENT
STEROID HORMONE RECEPTOR ACTIVITY

Glutamate

iGSEA4GWAS

GLUTAMATE RECEPTOR ACTIVITY
G PROTEIN SIGNALING ADENYLATE CYCLASE INHIBITING PATHWAY
GLUTAMATE SIGNALING PATHWAY
NEUROTRANSMITTER SECRETION
REGULATED SECRETORY PATHWAY
METABOTROPIC GLUTAMATE GABA B LIKE RECEPTOR ACTIVITY
SYNAPTIC VESICLE

GSA-SNP

GLUTAMATE RECEPTOR ACTIVITY
GLUTAMATE RECEPTOR BINDING
GLUTAMATE RECEPTOR SIGNALING PATHWAY
REGULATION OF NEUROTRANSMITTER SECRETION
NEUROPEPTIDE SIGNALING PATHWAY
POSITIVE REGULATION OF SYNAPTIC TRANSMISSION
POSITIVE REGULATION OF TRANSMISSION OF NERVE IMPULSE
SYNAPTIC VESICLE
SYNAPSE
SYNAPSE ORGANIZATION
SYNAPSE PART
SYNAPTIC MEMBRANE

Immunity

IGSEA4GWAS

ACTIVATION OF IMMUNE RESPONSE
HSA04612 ANTIGEN PROCESSING AND PRESENTATION
HSA04940 TYPE I DIABETES MELLITUS
IMMUNOLOGICAL SYNAPSE
INTERLEUKIN 8 BIOSYNTHETIC PROCESS
INTERLEUKIN 8 PRODUCTION
DEFENSE RESPONSE
INFLAMMATORY RESPONSE
RESPONSE TO STRESS
IMMUNE SYSTEM PROCESS
IMMUNE RESPONSE
REGULATION OF RESPONSE TO STIMULUS
DEFENSE RESPONSE TO VIRUS
TRANSMEMBRANE RECEPTOR PROTEIN PHOSPHATASE ACTIVITY
EPA4PATHWAY
TCRAPATHWAY

GSA-SNP

ACTIVATION OF IMMUNE RESPONSE
HSA04612 ANTIGEN PROCESSING AND PRESENTATION
HSA04940 TYPE I DIABETES MELLITUS
IMMUNE RESPONSE ACTIVATING CELL SURFACE RECEPTOR SIGNALING PATHWAY
IMMUNE RESPONSE ACTIVATING SIGNAL TRANSDUCTION
IMMUNE RESPONSE REGULATING CELL SURFACE RECEPTOR SIGNALING PATHWAY
IMMUNE RESPONSE REGULATING SIGNALING PATHWAY
IMMUNE SYSTEM DEVELOPMENT
POSITIVE REGULATION OF IMMUNE RESPONSE
POSITIVE REGULATION OF IMMUNE SYSTEM PROCESS
REGULATION OF IMMUNE RESPONSE
ANTIGEN PROCESSING AND PRESENTATION OF PEPTIDE OR POLYSACCHARIDE ANTIGEN VIA
ANTIGEN RECEPTOR MEDIATED SIGNALING PATHWAY
INTERFERON GAMMA MEDIATED SIGNALING PATHWAY
RESPONSE TO INTERFERON GAMMA
T CELL RECEPTOR SIGNALING PATHWAY
MHC CLASS II PROTEIN COMPLEX
MHC CLASS II RECEPTOR ACTIVITY
MHC PROTEIN COMPLEX
REGULATION OF COMPLEMENT ACTIVATION

Adhesion/Proliferation

IGSEA4GWAS

HSA04510 FOCAL ADHESION
HSA04514 CELL ADHESION MOLECULES
HSA04530 TIGHT JUNCTION
CELL CELL ADHESION
REGULATION OF CELL ADHESION
ACTIN CYTOSKELETON
HSA05218 MELANOMA
HSA05216 THYROID CANCER
CYTOSKELETON
CELL DIVISION
CYTOKINESIS
NUCLEOTIDE KINASE ACTIVITY
ANCHORED TO PLASMA MEMBRANE
ANCHORED TO MEMBRANE
REGULATION OF CELL PROLIFERATION
NEGATIVE REGULATION OF CELL CYCLE
NEGATIVE REGULATION OF CELL ADHESION
EPIDERMAL GROWTH FACTOR RECEPTOR SIGNALING PATHWAY
POSITIVE REGULATION OF CELL MIGRATION
REGULATION OF CELL MIGRATION

GSA-SNP

HSA04510 FOCAL ADHESION
HSA04514 CELL ADHESION MOLECULES
HSA04530 TIGHT JUNCTION
CELL CELL ADHESION
REGULATION OF CELL ADHESION
ACTIN CYTOSKELETON
ACTIN BINDING
ACTIN CYTOSKELETON
ACTIN CYTOSKELETON ORGANIZATION
ACTIN FILAMENT BINDING
ACTIN FILAMENT BASED PROCESS
ADHERENS JUNCTION
ADHERENS JUNCTION ORGANIZATION
ANCHORING JUNCTION
CELL ADHESION MEDIATED BY INTEGRIN
INTEGRIN BINDING
INTEGRIN COMPLEX
INTEGRIN MEDIATED SIGNALING PATHWAY
CELL ADHESION MOLECULE BINDING
CELL CELL ADHERENS JUNCTION
CELL CELL ADHESION
CELL CELL JUNCTION
CELL CELL JUNCTION ORGANIZATION
CELL MATRIX ADHESION
CELL SUBSTRATE ADHERENS JUNCTION
CELL SUBSTRATE ADHESION
CELL SUBSTRATE JUNCTION
CELL SUBSTRATE JUNCTION ASSEMBLY
NEURON CELL CELL ADHESION

POSITIVE REGULATION OF CELL ADHESION

REGULATION OF CELL CELL ADHESION

REGULATION OF CELL MATRIX ADHESION

REGULATION OF CELL SUBSTRATE ADHESION

NEURON MIGRATION

POSITIVE REGULATION OF FIBROBLAST MIGRATION

POSITIVE REGULATION OF SMOOTH MUSCLE CELL MIGRATION

POSITIVE REGULATION OF SMOOTH MUSCLE CELL PROLIFERATION

REGULATION OF EPITHELIAL CELL PROLIFERATION

KERATINOCYTE PROLIFERATION

REGULATION OF CELL MIGRATION

POSITIVE REGULATION OF CELL MIGRATION

Development

iGSEA4GWAS

TISSUE MORPHOGENESIS
GLAND DEVELOPMENT
AXON GUIDANCE
HSA04340 HEDGEHOG SIGNALING PATHWAY
NEGATIVE REGULATION OF CELL DIFFERENTIATION
NEGATIVE REGULATION OF DEVELOPMENTAL PROCESS
ANATOMICAL STRUCTURE MORPHOGENESIS
ANATOMICAL STRUCTURE FORMATION
REGULATION OF ANGIOGENESIS
NEUROGENESIS
GENERATION OF NEURONS
NEURON DIFFERENTIATION
AXON GUIDANCE
CENTRAL NERVOUS SYSTEM DEVELOPMENT
NERVOUS SYSTEM DEVELOPMENT
ECTODERM DEVELOPMENT
EPIDERMIS DEVELOPMENT

GSA-SNP

TISSUE MORPHOGENESIS
GLAND DEVELOPMENT
AXON GUIDANCE
BRANCHING MORPHOGENESIS OF A TUBE
EPITHELIAL TUBE BRANCHING INVOLVED IN LUNG MORPHOGENESIS
EPITHELIAL TUBE MORPHOGENESIS
EPITHELIUM DEVELOPMENT
MORPHOGENESIS OF A BRANCHING EPITHELIUM
MORPHOGENESIS OF A BRANCHING STRUCTURE
MORPHOGENESIS OF AN EPITHELIUM
BLOOD VESSEL DEVELOPMENT
BLOOD VESSEL MORPHOGENESIS
BRAIN DEVELOPMENT
BRANCHING INVOLVED IN URETERIC BUD MORPHOGENESIS
LIMB DEVELOPMENT
LIMB MORPHOGENESIS
REGULATION OF CELL MORPHOGENESIS
REGULATION OF CELL MORPHOGENESIS INVOLVED IN DIFFERENTIATION
REGULATION OF DEVELOPMENTAL GROWTH
REGULATION OF MORPHOGENESIS OF A BRANCHING STRUCTURE
HEART DEVELOPMENT
MUSCLE ORGAN DEVELOPMENT
REGULATION OF ORGAN MORPHOGENESIS
REGULATION OF NERVOUS SYSTEM DEVELOPMENT
REGULATION OF NEUROGENESIS
RENAL SYSTEM DEVELOPMENT
RENAL SYSTEM PROCESS
RESPIRATORY SYSTEM DEVELOPMENT
RESPIRATORY TUBE DEVELOPMENT

SKELETAL SYSTEM DEVELOPMENT

TUBE DEVELOPMENT

TUBE MORPHOGENESIS

URETERIC BUD MORPHOGENESIS

VASCULATURE DEVELOPMENT

ENDOTHELIAL CELL DEVELOPMENT

ENDOTHELIUM DEVELOPMENT

EPITHELIUM DEVELOPMENT

LUNG DEVELOPMENT

LUNG MORPHOGENESIS