

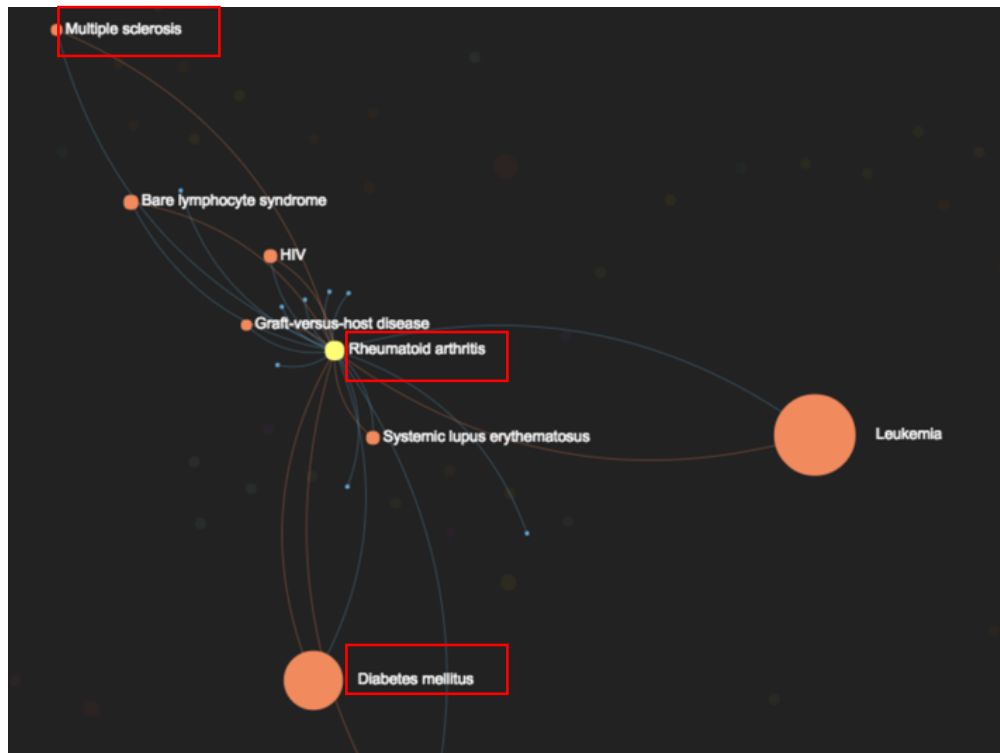
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Supplemental Data

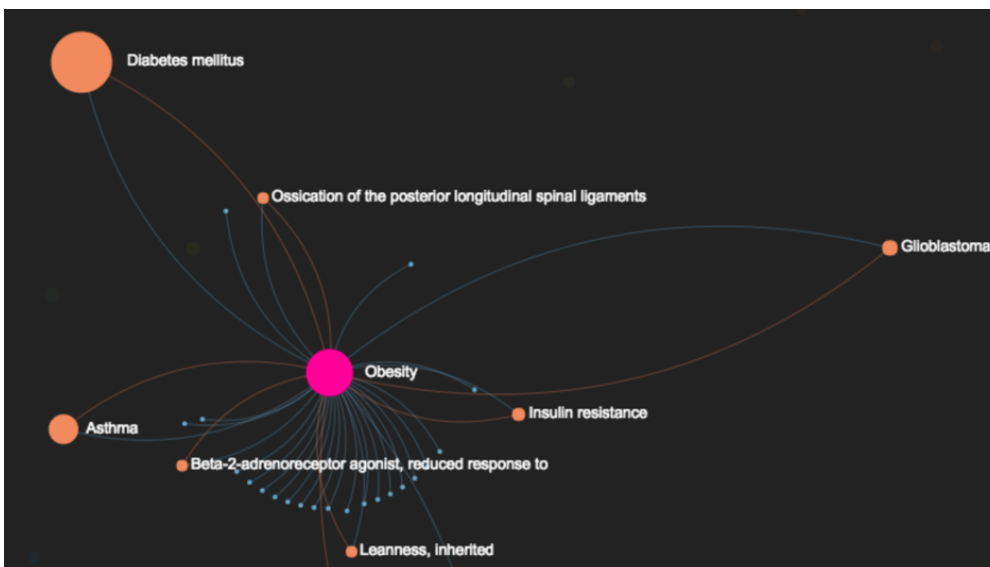
Human-Disease Phenotype Map Derived from PheWAS across 38,682 Individuals

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Supplementary Information



(A)



(B)

Figure S1. Network similarities with human disease network. (A) The disease connection between the three highlighted autoimmune diseases (Red box) in HDN are same as observed in the DDN. (B) Obesity and its neighboring diseases in HDN and the highlighted red box represent the connections found in our network as well.

Gene	Diseases	Shared SNPs	Known GWAS Associations	Enhancer Region Variants	Tissues
PTPN22	Type 1 Diabetes Multiple sclerosis	1	Previously Known	exm85427	Breast Dnd41_TCell_Leukemia
BAG6	Type 1 Diabetes Multiple sclerosis	1	Previously Known	rs760293	Adrenal HepG2_Hepatocellular_Carcinoma Liver
BTNL2	Type 1 Diabetes Rheumatoid arthritis	2	Previously Known	rs3129953	Breast Cervix
C2	Type 1 Diabetes Multiple sclerosis	1	Novel	N/A	N/A
C6orf10	Type 1 Diabetes Multiple sclerosis Rheumatoid arthritis	4	Previously Known	N/A	N/A
CFB	Type 1 Diabetes Multiple sclerosis	1	Previously Known	rs1048709	Pancreas Placenta
HCG20	Type 1 Diabetes Rheumatoid arthritis	1	Previously Known	rs6920124	Blood Bone Cervix Dnd41_TCell_Leukemia GI Smooth Muscle HepG2_Hepatocellular_Carcinoma Stromal Connective Stem cells Thymus
HCG26	Type 1 Diabetes Multiple sclerosis	1	Novel	rs2523663	Blood Heart Spleen
HLA-DOB	Type 1 Diabetes Multiple sclerosis	1	Previously Known	rs2071469	Placenta Stromal Connective Stem cells
HLA-DQA1	Type 1 Diabetes Multiple sclerosis	1	Novel	N/A	N/A
HLA-DQA2	Type 1 Diabetes Multiple sclerosis	1	Previously Known	N/A	N/A
HLA-DQB1	Type 1 Diabetes Multiple sclerosis	1	Novel	N/A	N/A
HLA-DRA	Type 1 Diabetes Multiple sclerosis	1	Previously Known	N/A	N/A
LOC101929072	Type 1 Diabetes Rheumatoid arthritis	1	Previously Known	rs2251396	A549_EtOH_0.02pct_Lung_Carcinoma Blood Bone Breast Dnd41_TCell_Leukemia Fat Skin (Adipose Tissue)
LOC102725019	Type 1 Diabetes Multiple sclerosis	1	Previously Known	N/A	N/A
NOTCH4	Type 1 Diabetes Multiple sclerosis	2	Previously Known	N/A	N/A
PSMB8	Type 1 Diabetes Multiple sclerosis	1	Novel	N/A	N/A
TAP1	Type 1 Diabetes Multiple sclerosis	1	Previously Known	rs3198005	Breast Cervix
TAP2	Type 1 Diabetes Multiple sclerosis Rheumatoid arthritis	5	Previously Known	rs3819721 rs241426 rs3819714	Blood Breast Dnd41_TCell_Leukemia HepG2_Hepatocellular_Carcinoma Placenta Thymus Cervix

Table S2. Shared SNPs between disease network. Here we present all the SNPs shared between key hub nodes in the network i.e. Type 1 Diabetes, Multiple Sclerosis, and Rheumatoid Arthritis.

Gene ontology term	p-value	FDR q-value
Antigen processing and presentation ion of peptide antigen	9.74e ⁻¹⁶	5.76e ⁻¹²
MHC class II receptor activity	2.87e ⁻¹⁵	6.47e ⁻¹²
Immune system process	3.53e ⁻¹⁵	6.47e ⁻¹²
Antigen processing and presentation	4.38e ⁻¹⁵	6.47e ⁻¹²
MHC class II protein complex	1.58e ⁻¹⁴	1.87e ⁻¹¹
Positive regulation of immune system process	9.36e ⁻¹⁴	9.23e ⁻¹¹
Regulation of immune system process	2.16e ⁻¹³	1.83e ⁻¹⁰
MHC protein complex	2.91e ⁻¹³	2.15e ⁻¹⁰
Immune response	9.9e ⁻¹³	6.51e ⁻¹⁰
Regulation of immune resonance	5.62e ⁻¹²	3.33e ⁻⁹

Table S3. Gene Ontology enrichment analysis using shared genes between Type 1 Diabetes, Multiple Sclerosis, and Rheumatoid Arthritis. P-values were obtained from the hypergeometric distribution