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Supplemental Data

**PheWAS and Beyond: The Landscape of Associations
with Medical Diagnoses and Clinical Measures
across 38,662 Individuals from Geisinger**

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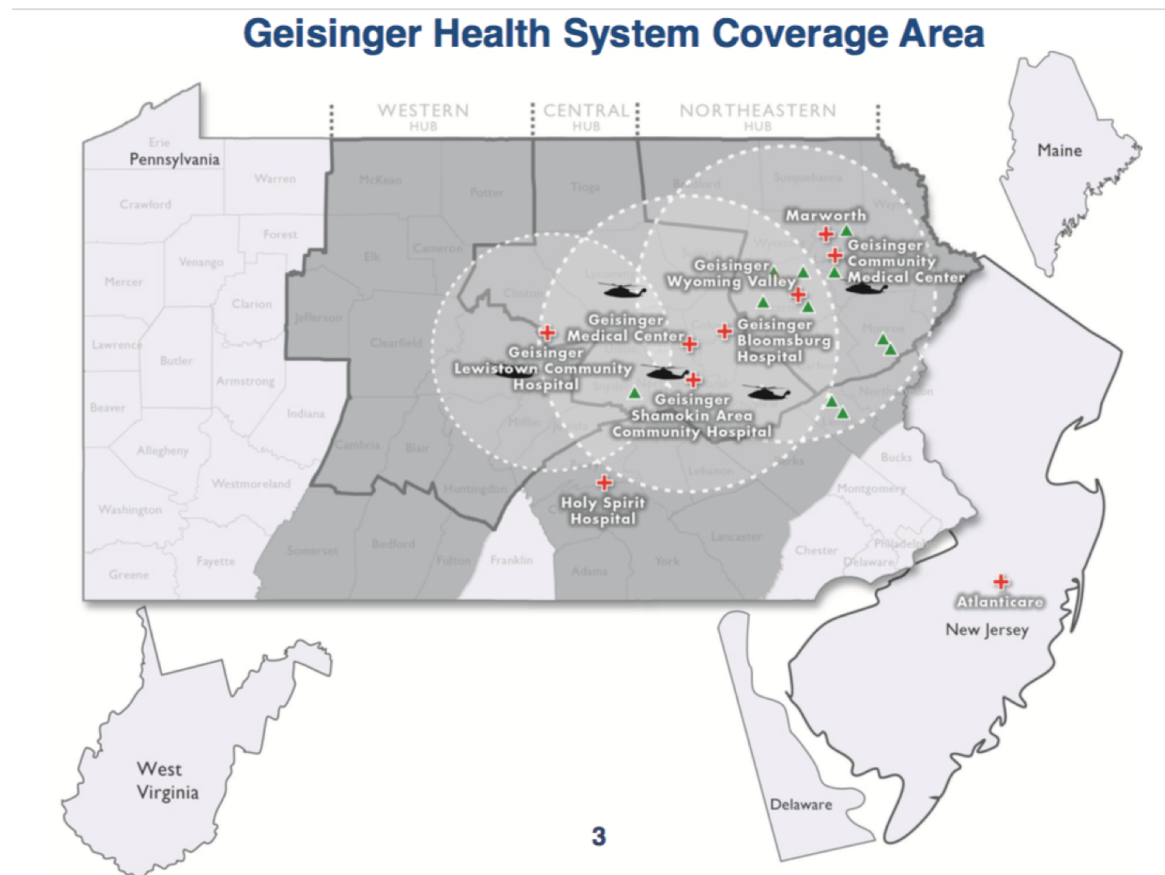


Figure S1. Geisinger Health System. Geisinger health system headquartered in Danville, Pennsylvania. The heat map shows the density of patients across different counties in the state of Pennsylvania, who have enrolled Geisinger as primary care.

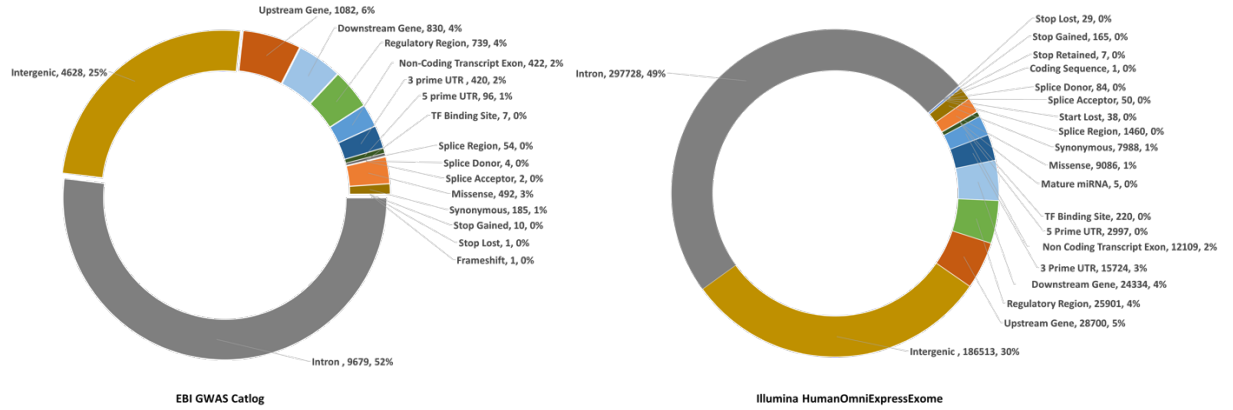


Figure S2. Comparison of VEP annotations. In the plot, we show the side by side comparison of the VEP annotations between SNPs from EBI GWAS catalog and the genotyped variants from identified in our study population.

Table S1. Summary of 25 clinical lab measures included in the study.

Phenotype	Transformation	Mean	Median	Min	Max
Alanine Aminotransferase	Natural Log	3.11	3.09	1.10	4.34
Albumin	Natural Log	1.44	1.46	0.59	1.69
Alkaline Phosphatase	Natural Log	4.30	4.29	2.40	5.16
Anion Gap	-	10.09	10.00	5.50	14.50
Aspartate Aminotransferase	Natural Log	3.17	3.14	1.87	4.17
Bilirubin	Natural Log	-0.84	-0.91	-6.91	0.53
Calcium	-	9.40	9.40	8.55	10.25
Carbon dioxide	-	26.91	27.00	21.70	32.15
Chloride	-	102.60	103.00	97.00	108.00
Creatinine	Natural Log	-0.17	-0.22	-2.30	0.70
Erythrocyte Distribution width (RDW)	Natural Log	2.60	2.59	2.41	2.80
Glucose	Natural Log	4.63	4.58	3.43	5.26
Hematocrit	-	39.97	40.00	30.30	49.45
Hemoglobin	-	13.52	13.50	9.95	17.05
Mean Corpuscular Hemoglobin Concentration	-	33.83	33.90	31.55	36.05
Mean Corpuscular Hemoglobin	-	30.32	30.40	25.65	34.85
Mean Corpuscular Volume	-	89.57	89.60	77.70	101.25
Platelet Count	-	241.00	238.00	83.50	404.00
Platelet Mean Volume	-	9.76	9.80	6.85	12.65
Potassium	-	4.25	4.25	3.55	4.95
Protein	-	6.99	7.00	5.95	8.00
RBC Count	-	4.49	4.49	3.32	5.63
Sodium	-	139.60	140.00	135.00	144.00

Urea Nitrogen	-	15.74	15.00	2.00	32.50
WBC Count	Natural Log	1.99	1.99	-6.91	5.99

Table S3. Fine mapping chromosome using functional annotations. The associations of variants in LD haplotype block on chromosome 11 and its functional annotations with most-probable chromatin state and gene correlation from RNA-Seq data.

Phenotype	SNP	P-Value	Gene	R ²	Chromatin State (Most Probable)
Psoriasis (696.1)	rs9501032	1.4 x 10 ⁻¹¹	DDR1	0.45	Enhancer
Hypercoagulable state (289.81)	rs2038024	4.1 x 10 ⁻³⁰	RP1-206D15.5	0.17	Active TSS
Essential Hypertension (401.9)	rs964184	9.13 x 10 ⁻⁵	APOA1-AS	0.22	Strong Transcription
Coronary Atherosclerosis (414.00)	rs964184	6.73 x 10 ⁻⁵	APOA1-AS	0.22	Strong Transcription
Pure Hyperglyceridemia (272.1)	rs964184	6.3 x 10 ⁻¹⁵	APOA1-AS	0.22	Strong Transcription
Hyperlipidemia (272.4)	rs964184	4.66 x 10 ⁻⁸	APOA1-AS	0.22	Strong Transcription