SUPPLEMENTARY DATA

Supplementary Table 1. Lipodystrophy phenotype query and comorbidities code list:

Condition	ICD-9-CM	ICD-10-CM
Lipodystrophy	272.6	E88.1
Hyperlipidemia	272	E78
Diabetes	250	E11
Hypertension	401	I10
Non-alcoholic fatty liver	571.8	K75.8, K76
HIV	042	B20

SUPPLEMENTARY DATA

Supplementary Table 2. Observed prevalence of lipodystrophy/lipoatrophy in MarketScan database and age-sex-standardized prevalence projected to the US population on July 01, 2017

Sex	Age group, years	N of prevalent cases in MarketScan	N of enrollees in MarketScan	Estimated prevalence in MarketScan (per million)	US population	Estimated prevalence in the US (counts)
Male	18-34	107	13,823,153	7.7	38,688,405	299
Male	35-49	164	11,014,972	14.9	30,755,275	458
Male	50-64	401	10,956,355	36.6	30,777,985	1,126
Male	65-79	199	4,428,722	44.9	17,775,956	799
Male	80+	50	1,059,709	47.2	4,788,728	226
Female	18-34	277	14,042,227	19.7	37,270,736	735
Female	35-49	1,125	11,846,952	95.0	31,093,953	2,953
Female	50-64	1,256	12,048,096	104.2	32,618,767	3,400
Female	65-79	392	4,941,843	79.3	20,648,751	1,638
Female	80+	58	1,526,167	38.0	7,645,244	291
Total		4,029	85,688,196		252,063,800	11,925
Prevalence		47.0/million (crude)			47.3/million (age-sex-standardized)	

Lipodystrophy cases defined as individuals with one inpatient or two outpatient diagnosis code of ICD-9 272.6/ICD-10 E88.1.

SUPPLEMENTARY DATA

Supplementary Table 3. Lipodystrophy associated variants identified in DiscovEHR cohort.

CHR	POSITION	REF	ALT	GENE	ANNOTATION	NT_CHANGE	AA_CHANGE	CLINVAR_ID	EXAC_FREQ	CLASSIFICATION
1	156136985	G	A	*LMNA*	missense	c.G1445A	p.R482Q	VCV000014486	8.32E-06	Pathogenic
1	156138534	G	A	*LMNA*	missense	c.G1745A	p.R582H	VCV000014494	0	Pathogenic
3	12392713	C	T	*PPARG*	missense	c.C496T	p.R166W	VCV000008144	na	Pathogenic
3	12416836	G	A	*PPARG*	missense	c.G868A	p.V290M	VCV000008137	8.24E-06	Pathogenic
5	68296248	G	A	*PIK3R1*	missense	c.G992A	p.R331Q	VCV000126459	na	Pathogenic
5	68296301	C	T	*PIK3R1*	missense	c.C1045T	p.R349W	VCV000060763	na	Pathogenic
3	12392714	G	A	*PPARG*	missense	c.G497A	p.R166Q	VCV000436397	na	Likely_Pathogenic
3	12379922	G	T	*PPARG*	nonsense	c.G217T	p.E73X	na	na	Expected_Pathogenic
3	12379932	G	C	*PPARG*	splicing	c.226+1G>C	.	na	na	Expected_Pathogenic
3	12405881	G	A	*PPARG*	splicing	c.536-1G>A	.	na	na	Expected_Pathogenic